**Universidade de Aveiro** Secção Autónoma de Ciências da Saúde
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**Álvaro Filipe Ribeiro dos Santos Oliveira Mendes**

# Família e aconselhamento genético: o caso dos cancros hereditários

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Tese apresentada à Universidade de Aveiro para cumprimento dos requisitos necessários à obtenção do grau de Doutor em Ciências e Tecnologias da Saúde, realizada sob a orientação científica da Doutora Liliana Xavier Marques de Sousa, Professora Auxiliar com Agregação na Secção Autónoma de Ciências da Saúde da Universidade de Aveiro.

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Dedico este trabalho à minha família.

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**palavras-chave**

Família; aconselhamento genético; cancros hereditários; genética psicossocial; grupos de discussão multifamílias; metodologia qualitativa.

**resumo**

Os desenvolvimentos associados à medicina genómica e biologia molecular representam novas possibilidades no diagnóstico, tratamento e prevenção de doenças comuns, confrontando indivíduos e famílias com complexos desafios à integração da informação genética na gestão da saúde e nas suas vidas. Este estudo centra-se em famílias com suscetibilidade genética acrescida a cancros hereditários e pretende contribuir para o conhecimento da experiência individual e familiar do aconselhamento oncogenético, incluindo como pode ser contemplada no desenvolvimento de intervenções de apoio psicossocial e na organização dos cuidados de saúde na era (pós)genómica. O processo de investigação incorpora perspetivas da genética psicossocial e da teoria dos sistemas familiares. Engloba metodologias qualitativas de recolha e análise de dados, envolvendo indivíduos, famílias e profissionais de saúde num formato de investigação-ação participativa.

Os principais resultados permitem: i) conceptualizar a experiência do aconselhamento oncogenético, através da caracterização das suas implicações instrumentais, emocionais, relacionais e desenvolvimentais para o indivíduo e sistema familiar; ii) conhecer o desenvolvimento, implementação e avaliação de um programa psicoeducativo multifamiliar, enquanto intervenção de apoio psicossocial a indivíduos com suscetibilidade acrescida a cancros hereditários e suas famílias; e iii) integrar a perspetiva dos profissionais de saúde quanto à incorporação de apoio psicossocial na provisão dos serviços oncogenéticos. As conclusões gerais sustentam a importância do aprofundamento da pesquisa sobre o funcionamento familiar face ao aconselhamento e risco oncogenético, e a incorporação de uma orientação familiar nesses serviços.

As implicações decorrentes da suscetibilidade acrescida a doenças genéticas impõem uma discussão alargada aos vários agentes envolvidos no planeamento, provisão e utilização dos cuidados de saúde, no sentido do desenvolvimento de serviços atuantes no *continuum* biopsicossocial indivíduo-família-sistema de saúde-comunidade.

**keywords**

Family; genetic counselling; hereditary cancers; psychosocial genetics; multifamily discussion groups; qualitative methodology.

**abstract**

As genomic medicine and molecular biology expands, new possibilities for diagnosis, treatment and prevention of common diseases are increasingly available, confronting individuals and families with complex challenges regarding the integration of this new information in health management and into their lives.

This thesis focuses on families with increased genetic susceptibility for hereditary cancers. It aims to contribute to a deeper understanding of the individual and familial experience of oncogenetic counselling, and how it can be considered for developing adequate psychosocial support and on the provision of healthcare services in the (post)genomics era. The research process draws on perspectives from psychosocial genetics and family systems theory, enfolding qualitative research methods of collection and data analysis, and involving individuals, families and genetics healthcare professionals in a participatory action-research design.

Main findings were: i) the conceptualization of the oncogenetic counselling experience, addressing its instrumental, emotional, relational, and developmental implications for the individual and the family system; ii) the planning, implementation, and evaluation of a multifamily psychoeducational intervention as a psychosocial tool for supporting cancer susceptibility families; and iii) the description of the professionals' views on the incorporation of psychosocial support in the provision of oncogenetic counselling services. General conclusions emphasize the need to promote further family-oriented research in oncogenetic counselling, and to acknowledge and incorporate these data as part of service delivery.

Acknowledging the implications derived from genetic risk call for a broad participation of the diverse stakeholders involved in genetics healthcare, from policy-makers to providers and users, towards the provision of effective services on the biopsychosocial *continuum* individual-family-healthcare system-community.

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# INTRODUÇÃO

## DO LABORATÓRIO À FAMÍLIA: CONTEXTO SISTÉMICO DAS DOENÇAS GENÉTICAS

Entre 5 a 15% dos casos de cancro envolvem a transmissão hereditária de mutações genéticas. Ao longo das duas últimas décadas, os avanços tecnocientíficos no campo da biologia molecular permitiram aprofundar o estudo dos genes na predisposição a várias síndromes oncológicas, resultando na crescente disponibilização de testes genéticos. Estes determinam, com precisão variável, o risco individual de desenvolvimento da doença, num processo com implicações para além do domínio biomédico e individual. O recurso a testes genéticos e a sua incorporação nos serviços clínicos materializa a era genómica dos cuidados em saúde.

O projeto inicialmente submetido e aprovado pela FCT centrava-se no desenvolvimento, implementação e avaliação de uma intervenção psicoeducativa para indivíduos e familiares no contexto do aconselhamento oncogenético em casos de cancro colorectal. Porém, desde cedo foram introduzidas modificações ao plano inicial, nomeadamente, a expansão do estudo a outros tipos de cancros hereditários, como o cancro da mama e ovários, cuja prevalência e implicações clínicas e psicossociais assumem relevância no contexto do risco oncogenético. Esta tese procura ainda compreender a experiência individual e familiar do aconselhamento oncogenético. Para tal, focámo-nos em indivíduos portadores de mutações de predisposição oncogenética conhecidas e suas famílias. Pretendemos também, por outro lado, alargar o enfoque da investigação até aos profissionais de saúde dos serviços de genética, de forma a abranger os intervenientes do sistema em interação: indivíduo, família e serviços de saúde.

Sendo esta tese composta por um conjunto de estudos apresentados sob a forma de artigos científicos, publicados ou em vias de publicação, a introdução que se segue contextualiza teoricamente a interligação e evolução dos mesmos, enquadrando os principais referenciais teóricos da tese e enunciando o seu enfoque e objetivos. Assim, o enquadramento teórico apresenta a *nova genética* a partir da crescente incorporação do conhecimento tecnocientífico na saúde (Clarke, Shim, Fosket, & Nelson, 2009)[1], e da genética psicossocial, que procura estudar as suas implicações ultrapassando uma perspetiva estritamente biomédica (Harper, 1993). A caracterização do aconselhamento genético e dos aspetos clínicos dos cancros hereditários são igualmente destacados, bem como o impacto das doenças genéticas na família através de uma "lente" compreensiva sistémica (Miller, McDaniel, Rolland, & Feetham, 2006).

## 1. ÂMBITO DA *NOVA GENÉTICA*

Em abril de 2003, 50 anos após James Watson e Francis Crick terem identificado a estrutura da molécula do ADN[2], e assim inaugurado a genética molecular, foi concluído o mapeamento e sequenciação dos cerca de três biliões de pares de bases do ADN que formam o genoma humano. Esta descoberta, através do Projeto do Genoma Humano[3], confirmou o que se antecipara como a "nova era" da genómica, definida como "*(…) the study of the entire human genome; as applied to health outcomes, genomic research includes interactions of multiple genes with each other and with the environment*" (Hay *et al*., 2007: 276). A investigação genómica avançada permitiu a identificação e clonagem do(s) gene(s) associado(s) a diversas doenças comuns (como os cancros, por exemplo) e doenças genéticas raras, reconhecendo a(s) mutação(ões) implicada(s) e sua localização. Também permitiu a criação de testes de ADN que diagnosticam a presença dessas mutações genéticas em indivíduos assintomáticos, predizendo maior suscetibilidade de desenvolverem a doença no futuro (Collins, Green, Guttmacher, & Guyer, 2003). Os testes genéticos permitem aos utilizadores o acesso a informação preditiva que implica decisões para a

[1] A referenciação bibliográfica usada respeita os critérios utilizados nas publicações às quais os artigos foram submetidos. Como tal, o leitor encontrará discrepâncias neste aspeto ao longo da tese. Na introdução e conclusões gerais, a referenciação bibliográfica segue o estilo da Associação Americana de Psicologia (*American Psychological Association*, APA).

[2] O ADN (Ácido Desoxirribonucleico) é o nome químico da molécula que codifica a informação genética contida em todos os organismos vivos. É uma cadeia de milhares de nucleótidos com duas bandas ligadas em hélice por elos de hidrogénio entre pares de bases azotadas (entre adenina e timina e entre guanina e citosina) (Hodgson, Foulkes, Eng, & Maher, 2007).

[3] O Projeto do Genoma Humano (*Human Genome* Project) é um projeto científico internacional iniciado em outubro de 1990 por instituições governamentais dos Estados Unidos da América (*National Institute of Health* e *Department of Energy*) e mais tarde apoiado política, institucional e financeiramente por vários países, entre os quais o Japão, França, Reino-Unido e Alemanha (Collins, Green, Guttmacher, & Guyer, 2003).

gestão da saúde individual e familiar, com forte impacto emocional e psicossocial a curto, médio e longo prazo (Evers-Kiebooms, Welkenhuysen, Claes, Decruyenaere, & Denayer, 2000).

As referências ao termo *nova genética* remontam aos anos 1970, aquando do advento da engenharia genética e, nomeadamente, da tecnologia associada ao ADN recombinante. Essa designação antecipava, na ótica da saúde pública, as implicações oriundas dos avanços da genética hoje disponíveis para o diagnóstico, tratamento e prevenção das doenças comuns (Petersen, 2002a). O termo foi definido e o seu campo de atuação amplamente debatido sobretudo durante a última década do século XX, quando os ecos do Projeto do Genoma Humano agitavam clínicos e investigadores provenientes de várias áreas do saber. A antropóloga Katja Finkler define deste modo a nova genética (2000: 50):

> "*The new genetics refers to knowledge and procedures based on DNA technology; it brings into the forefront of people's consciousness the genes carried by individuals and their families. It anticipates that by seeking testing, people will develop control over genetically inherited diseases. By doing so they might be enabled to plan their lives to the point of opting to bear only healthy children with predetermined characteristics"*.

A nova genética enfatiza o impacto da genética no modo como os indivíduos pensam e agem sobre a sua saúde, com repercussões na conceção do *self* e identidade que se estendem a "novas" formas de conceber a família, a saúde e a doença. Esses avanços situam o contexto clínico e de investigação da genética e genómica num complexo interface científico e social quanto à delimitação e assimilação das suas aplicações, significados e repercussões. Tornou-se consensual entre investigadores, sobretudo oriundos das ciências sociais, uma posição reflexiva face à produção e uso do conhecimento científico e à exploração das respostas do público à sua utilização. No espectro alargado da compreensão pública da ciência, a ciência médica, em particular, ocupou historicamente uma posição de relevo face a outros ramos científicos, angariando consistente atenção dos *media* e contribuindo para que as suas representações populares fossem permeadas por uma visão utilitarista (Durant, Hansen, & Bauer, 1996; Lambert & Rose, 1996). A genética é disso um bom exemplo, transportando para o debate público os avanços médicos que decorrem da biotecnologia (novas formas de diagnóstico e terapêutica) e respetivas considerações éticas, sociais e legais (Nelkin & Lindee, 2004).

Por entre discursos científicos, filosóficos, religiosos, políticos e institucionais e a visibilidade mediática, a esfera pública incorporou igualmente aspetos proto-políticos associados ao desenvolvimento genómico. Espaços participativos e movimentos sociais emergiram fora das estruturas institucionais da democracia representativa, acompanhando iniciativas de governança,

isto é, envolvendo formas colaborativas e não-hierarquizadas de participação cívica que contribuem para reenquadrar os discursos dominantes sobre a ciência e tecnologia[4] (Evans, Plows, & Welsh, 2007). Os "factos" científicos da nova genética, em particular os decorrentes das implicações da pesquisa biotecnológica, são, assim, integrados nas contingências do quotidiano de indivíduos, famílias e profissionais de saúde. Bruno Latour (2004: 232, *in* Lock, Freeman, Sharples, & Lloyd, 2006: 278) sugere a necessidade de negociar socialmente a produção de conhecimento científico "para além do laboratório", através da "problematização reflexiva da ciência": *reality is not defined by matters of fact; matters of fact are not all that is given in experience; matters of fact are only very partial and very political renderings of matters of concern"*.

### 1.1. Medicina preditiva e *indivíduo geneticamente em risco*

Aos desenvolvimentos decorrentes do conhecimento da estrutura molecular do ADN e, consequentemente, do diagnóstico genético, associou-se um paradigma emergente nas ciências médicas, dirigido a indivíduos saudáveis: a medicina preditiva. Esta "*avalia o risco e indica as condições em que a doença pode surgir, permitindo evitar situações patogénicas; não constata a existência de uma anomalia, mas antes define a possibilidade ou probabilidade do seu aparecimento*" (Ruffié, 1994: 61).

A medicina preditiva combina conhecimentos do risco genético individual e dos fatores de risco externos (*e.g*. ambientais, comportamentais) para predizer e, em última instância, prevenir a emergência da doença. A sua ação de cariz antecipatório, baseada em premissas probabilísticas, traduziu-se na prática médica na transição de uma intervenção assente na identificação e supressão de sintomas, para uma abordagem orientada para a predição e gestão do risco (Petersen, 2002a). A predição de uma condição patológica em indivíduos assintomáticos conduziu a uma conceptualização da noção de doença qualitativamente distinta, até então alicerçada no paradigma médico preventivo (Novas & Rose, 2000). O risco genético emerge contido num espectro latente e probabilístico que possibilita que indivíduos possam ser identificados como estando em risco para consequências adversas, antes do aparecimento de sintomas ou sinais, podendo até permanecer assintomáticos para sempre (Temple, McLeod, Gallinger, & Wright, 2001). Surge, desta forma, uma nova espacialização das doenças, bem como distintas relações entre sintomas, sinais e doença, atualizando o que Foucault (1976) apelidou de *medicina da vigilância*, emergente na primeira metade do século XX, após o paradigma da medicina hospitalar ou "clínica". A noção de risco já

[4] De Vries (2008) antevê, no entanto, que a tendência em a ciência sair da dependência do estado acompanhando a globalização neo-liberal, introduzirá novos desafios ao debate público sobre o impacto societal da genómica: "*As a result, the broader social and political considerations that were discussed in the early stages of the debate still play a modest role at best. As far as technical issues are concerned, trust is put in the established procedures for self-regulation in the medical world. Ethical issues are turned into a private matter, thus effectively removing them from the public forum"* (De Vries, 2008: 7).

não incorpora o sintoma ou o sinal enquanto elemento discreto, mas passa a habitar o espaço extracorporal consubstanciado em "fatores de risco" que assinalam uma potencial doença futura; ocorre o que (Armstrong, 1995) apelidou de "patologização dos estados pré-sintomáticos" através da vigilância de um espaço ordinal no qual o indivíduo saudável pode tornar-se ainda mais saudável. O discurso médico preditivo situa agora a doença num contexto temporal mais amplo: tenta "transformar" o futuro através da modificação de atitudes e comportamentos no presente.

A prática do aconselhamento genético distancia-se de outros encontros médicos, pois encerra um contexto onde as noções de doença e normalidade são (re)definidas como forma de lidar com a incerteza inerente ao estatuto de risco (Sarangi, Bennert, Howell, e Clarke, 2005). Ou seja, foca questões prognósticas, em que o paciente interage com o discurso médico na construção do conhecimento acerca da sua constituição genética e sobre o risco de poder vir a ser afetado no futuro. A típica dicotomia normalidade *versus* patologia sucumbe à incorporação da incerteza no diagnóstico genético. Sarangi e colaboradores (2005: 156) postulam ainda que o contexto genómico incorpora mudanças profundas na relação entre o conhecimento do perito (*expert*) e o conhecimento popular (*lay*), acarretando incertezas epistemológicas e ontológicas por via da diluição e pluralização do conhecimento:

> *In contemporary societies, experts no longer solve problems through a straightforward application of theoretical / scientific knowledge, but increasingly engage in assessment of problems, and hence by extension, generation of risks associated with events that might or might not occur. In healthcare generally, and genetics in particular, we see that experts do not always possess the exact knowledge to explain in a strictly causal manner what one's risk status is or will become and what means there are, if any, for avoiding such risks"*

O contexto da nova genética encapsulou uma nova entidade médica: o *indivíduo geneticamente em risco* (Kenen, 1996). O modo como a medicina integrou o estatuto de risco na sua *praxis* potenciou o desenvolvimento de significativas alterações na relação entre clínico e paciente, no sentido de uma maior responsabilização dos indivíduos pelo controlo e gestão da sua saúde (Novas & Rose, 2000). O aconselhamento genético requer um papel proactivo e "proto-profissional" do paciente para a identificação da doença, pressupondo a sua atuação racional e prudente na gestão do risco. Dota o paciente de atributos informativos e processuais que o tornam "aliado" do clínico, incluindo a partilha da história médica pessoal e familiar, ou a tomada de decisões sobre o envolvimento em medidas profiláticas que podem reduzir o risco de ocorrência da doença. Inclui-se aqui igualmente: transmissão de informação genética relevante a familiares potencialmente em risco, crucial no âmbito alargado do planeamento da vida individual e familiar; vigilância e deteção precoces de

sinais da doença que permitam acompanhamento médico; tomada de decisões informadas sobre aspetos reprodutivos e gestão das carreiras profissionais (Kessler, 1979a). Kenen (1996) considera que o estatuto de *estar em risco* pressupõe, ainda, o desenvolvimento de uma relação simbiótica entre o indivíduo *em risco* e mecanismos tecnológicos relacionados com testes genéticos e monitorização do risco, envolvendo a criação de novas tarefas clínicas amplamente associadas ao uso da tecnologia na prática médica.

O contexto biopsicossocial do *indivíduo geneticamente em risco* contém novas práticas contemporâneas de identidade e subjetividade cujas implicações no modo de perspetivar o *self* e a vida envolvem o indivíduo e movimentos coletivos. Novas e Rose (2000) propuseram o termo *individualidade somática* para designar novas relações estabelecidas entre o corpo e o *self* do indivíduo em risco, e entre este e a comunidade: grupos, associações, comunidades de pessoas em risco, grupos de pacientes ou familiares ou participantes em ensaios clínicos de novas terapias génicas[5]. É conhecida a atividade de práticas individuais e coletivas ligados à saúde e genómica, que possibilitaram interpenetrações entre novos atores sociais, profissionais e governamentais, traduzindo novos modos de *biosocialidade* e *biocidadania* (Filipe, 2010; Gibbon, 2008; Nunes, 2006). O antropólogo Paul Rabinow formulou o termo *biosocialidade* para designar o efeito coletivo da genética e genómica a nível da organização social e da(s) sua(s) função(ões). Novas formas de identidade e subjetividade coletivas resultam, por exemplo, na abertura de novos espaços de participação no contexto dos cuidados de saúde para doentes e seus cuidadores e familiares[6] (Filipe, 2009; Nunes, Filipe, & Matias, 2008) e de ativismo transdisciplinar (Evans, Plows, & Welsh, 2007). A atividade das associações de doentes e de familiares de doentes enquanto atores coletivos na governação da saúde configuram redes (bio)sociais de indivíduos geneticamente em risco que emergem enquanto entidades comunitárias fora do âmbito profissional médico, podendo assumir igualmente o formato de grupos de suporte para pacientes, *fóruns* na *Internet*, ou coligações de organizações e grupos de autoajuda (Raz, 2010). De entre os objetivos destas "alianças genéticas" encontram-se a participação nos mecanismos decisórios públicos, juntamente

[5] A terapia génica consiste na transferência de uma cópia terapêutica ou funcional de um gene para uma localização inespecífica do genoma (isto é, de todo o material genético contido nos cromossomas) do indivíduo, de modo a reparar o gene deletério. Um novo gene, "normal", pode ser introduzido para substituir o gene disfuncional com o objetivo de curar ou modificar favoravelmente o curso clínico (Human Genome Project, 2011a).

[6] Existem trabalhos sobre a atividade das associações de doentes e de familiares de doentes enquanto atores coletivos na governação da saúde. McAllister, Dunn, e Todd (2010), por exemplo, incluíram representantes de associações de doentes no seu estudo sobre a validação e relevância do *empowerment* enquanto constructo individual qualitativo para avaliar os serviços de genética. Rabeharisoa (2006) descreveu o caso exemplar da Associação Francesa de Distrofia Muscular, que gere a sua política de investigação e envolve doentes e familiares como "peritos em experiências" através da sua ação enquanto parceiros dos especialistas médicos, produzindo em conjunto conhecimento sobre a doença, cuidados e terapias. Entre nós, a abertura de novos espaços de participação para doentes fora do âmbito profissional médico é ainda limitada, embora, como afirmam Nunes, Filipe e Matias (2008: 3), possam desempenhar um papel relevante no futuro: "*As associações de doentes promovem práticas inovadoras de mediação entre participantes heterogéneos no campo da saúde, como os profissionais e as instituições de prestação de cuidados, os governantes e decisores políticos, as comunidades científicas e de investigação, os prestadores de cuidados não-convencionais e a indústria farmacêutica*".

com os profissionais e outros agentes; o desenvolvimento de campanhas de literacia genética promotoras de decisões informadas sobre a utilização de serviços genéticos e biotecnológicos, bem como de direitos dos pacientes e de medidas antiestigmatização; e a comunicação junto dos profissionais de saúde das necessidades e recursos de indivíduos geneticamente em risco e suas famílias (Raz, 2010).

### 1.2. Biomedicalização da saúde, geneticização e emergência da genética psicossocial

Michel Foucault (1994: 145) empregou a noção de *biopoder* para traduzir "(…) *o que faz entrar a vida e os seus mecanismos no domínio dos cálculos explícitos e faz do poder-saber um agente de transformação da vida humana*". As suas formulações em torno das relações entre política, ciência e medicina (a "anátomo-política" da administração dos corpos e a "bio-política" da gestão da vida das populações) têm sido utilizadas para iluminar discussões em torno das implicações "produtivas" da nova genética. Ou seja, se o enfoque do risco, do ponto de vista sócio-antropológico, estende a jurisdição médica para além do corpo, a genética e a genómica incorporam uma mudança epistémica no sentido da *molecularização* do *olhar clínico*, envolvendo inovações tecnocientíficas socialmente percebidas de modo imperativo (Clarke *et al*., 2009). Por exemplo, os potenciais benefícios acoplados à investigação genómica sugerem a promessa de uma compreensão aprofundada, tratamento e prevenção de várias doenças, antecipada nas predições formuladas pelo Projeto do Genoma Humano (Human Genome Project, 2011b):

> "*Rapid and more specific diagnostic tests will make possible earlier treatment of countless maladies. Medical researchers also will be able to devise novel therapeutic regimens based on new classes of drugs, immunotherapy techniques, avoidance of environmental conditions that may trigger disease, and possible augmentation or even replacement of defective genes through gene therapy*".

As transformações associadas ao *biopoder* manifestam-se igualmente a nível societal. Adele Clarke e a sua equipa de colaboradores (Clarke, Shim, Mimo, Fosket, & Fishman, 2003) designaram por *biomedicalização* um conjunto de processos interativos que incluem a reformulação da economia política da saúde no sentido do desenvolvimento de sistemas de cuidados assentes no risco e vigilância e com enfoque na utilização de recursos tecnocientíficos. Os autores antecipam que o impacto decorrente da biomedicalização na produção, distribuição e consumo da medicina tem repercussões na gestão da informação médica, atribuindo novas propriedades ao "corpo" e produzindo novos atores e identidades individuais e coletivas.

A ação médica decorrente dos avanços tecnocientíficos é tida como prerrogativa para a atuação nos contextos genómico e pós-genómico (Clarke *et al*., 2009), no sentido da "política da saúde positiva" (Nunes, 2011). A sua potencial aplicação na otimização da saúde pressupõe indivíduos crescentemente responsáveis pela sua monitorização e prevenção, movidos por um imperativo ético-moral face ao uso do saber médico (Clarke *et al*., 2009). O desenvolvimento exponencial da tecnobiologia molecular tem sido acompanhado por intensa aclamação mediática, que ajuda a legitimar a biomedicina e o seu discurso científico (Lambert & Rose, 1996; Petersen, 2002b).

Estudos descrevem como o risco genético molda padrões decisórios e obrigações em famílias de indivíduos afetados ou em risco de doenças genéticas (Featherstone, Atkinson, Bharadwaj, & Clarke, 2006; Konrad, 2005; Rhodes, 1998). Fundados em abordagens etnográficas e recorrendo a métodos fenomenológicos, alguns autores oriundos das ciências sociais cunharam a ideia de "responsabilidade genética" ou "prudência genética" para descrever o caráter "obrigacionista" de os indivíduos estarem informados sobre a sua constituição genética, revelarem informação genética relevante aos familiares e adotarem medidas de vigilância e profiláticas para modelarem o seu risco (Kenen, 1996; Novas & Rose, 2000; Petersen & Lupton). Tais noções caracterizam identidades *geneticizadas* em termos individuais e formas de solidariedade genética extensivas aos familiares. No contexto da nova genética, a responsabilidade da gestão e prevenção do risco genético não é uma tarefa individual, mas, com efeito, algo sentido como uma "obrigação" ou "dever" familiar. Ou seja: os indivíduos não são somente responsáveis pela sua saúde, mas também pela dos outros (Hallowell, 1999; Lemke, 2007).

Vários autores exprimiram preocupações bioéticas face à crescente *geneticização* nos cuidados de saúde: Duster (1990) anteviu uma neo-eugenia perante um potencial reforço indireto de novas formas de racismo, de noções de superioridade ou inferioridade, desigualdades sociais e discriminações associadas à pesquisa do ADN; Lewontin, Rose, e Kamin (1986) e Nelkin e Tancredi (1994), por exemplo, alertaram para os perigos decorrentes de perspetivas reducionistas e deterministas da informação relativa ao património genético. Abby Lippman cunhou pela primeira vez o termo *geneticização* para ilustrar a sobrevalorização dos aspetos genéticos de uma dada situação em detrimento de outros, nomeadamente, de interpretações holísticas e de cariz social (1992: 1470):

> *"Geneticisation refers to an ongoing process by which (…) interventions employing genetic technologies are adopted to manage problems of health. Through this process, human biology is incorrectly equated with human genetics, implying that the latter acts alone to make us each the organism she or he is".*

Um aspeto central da geneticização é a assunção de um primado genético na explicação e na "construção" dos discursos sobre a saúde. Esta ideia consubstancia um *excepcionalismo genético* assente na crença de que os genes e o conhecimento genético / genómico adquirem precedência face a outros tipo de explicações ou de conhecimento e que desempenham, acima de que quaisquer outros fatores, um papel especialmente saliente na biologia e no comportamento humano. Alexandra Plows (2011) atribui a esta questão um padrão individualista de "olhar" a sociedade que tende a negligenciar explicações mais amplas e complexas para a expressão da influência dos genes na saúde. A autora relaciona as implicações da geneticização da saúde, identidade e cidadania com o determinismo genético, reducionismo e essencialismo; basicamente, essas noções sustentam a tese de que os genes operam de forma direta e inexorável na produção de evidências científicas e que a biologia humana, incluindo o comportamento e a identidade, são determinados "nos" genes[7]. Richards (2001) sugere que a "centração no gene" resulta do caráter unívoco associado às imagens culturais do ADN, fortemente conotadas com um estatuto "excecional", propondo a distinção entre "ADN biológico" e "ADN metafórico". Por outro lado, Clarke (1997a: 103) adverte para o facto de a individualização dos riscos genéticos tornar problemática a ação política da promoção da saúde pública para a população geral:

> "*Such a focus could lead to expansive, high-technology solutions to problems that might be better tackled by social or environmental means – but it would not lead to profits for the gene corporations or for the scientists who work in the blurred zone between academic research and the commercial applications of biotechnology"*.

Richards (1993) refletiu sobre a importância de as ciências sociais se debruçarem sobre as implicações da nova genética, atendendo ao seu teor vincadamente técnico e caráter obscuro da linguagem. Assistiu-se, assim, à emergência de uma nova área da genética médica: a genética psicossocial, centrada no estudo, em indivíduos e suas famílias, de preditores e processos relativos à complexa interconexão entre componentes psicológicas, sociais, relacionais e éticas dos testes

[7] Existem evidências que contradizem a ideia de que os genes materializam a sua ação através de uma trajetória mais ou menos pré-determinada ao longo da vida do indivíduo (Plows, 2011). Os recentes desenvolvimentos na biologia corroboram a tese de que os genes estão envolvidos de forma variável num intrincado sistema aberto e altamente complexo, juntamente com outros elementos biológicos. Pesquisas em torno dos sistemas de hereditariedade epigenética apontam para o caráter "moldável" da atividade dos genes e para a variabilidade da expressão génica (polimorfismos) em diferentes indivíduos, quer através da interação intergénica, quer de *inputs* ambientais (Jablonka & Lamb, 2005).
Naturalmente, os argumentos consistentes com a tese do *excepcionalismo genético* são válidos e legítimos. Com efeito, alguns exemplos sublinham a importância singular "do" gene: em casos de doenças monogénicas com penetrância completa; o facto de a informação genética possuir implicações para familiares devido aos padrões de transmissão hereditária através das gerações, desafiando, deste modo, as noções "tradicionais" da privacidade e confidencialidade individual (*cf*. pág. 22); ou de a informação genética poder identificar pessoas de forma rápida e sem conhecimento ou consentimento e de poder caracterizar indivíduos, espécies ou subgrupos da população.

preditivos e da prática do aconselhamento genético (Harper, 1993; Zagalo-Cardoso & Rolim, 2005).

A genética humana depara-se com desafios que têm exigido aturada atenção do ponto de vista bioético (Plows, 2011). Para além da já referida "ameaça neo-eugénica" e das derivações em torno da saúde e da identidade, do valor e da natureza humana, da justiça e equidade social, uma das áreas mais polarizadas do envolvimento público centra-se nas dimensões éticas subjacentes aos testes genéticos. Face à incorporação de mecanismos de genotipagem em serviços de sistemas de saúde, bem como de testes genéticos em programas de saúde pública e na prática clínica de vários países, alguns investigadores têm desenvolvolvido abordagens reflexivas quanto à produção de conhecimento em torno dos genes de suscetibilidade e criação de estimativas de risco para várias doenças "comuns" (como os cancros ou a doença de Alzheimer) (Lock *et al*., 2006). Assim, é na translação deste conhecimento científico para o "uso" quotidiano do tecido social que residem os desafios da genética psicossocial.

### 1.3. Os testes de suscetibilidade genética em perspetiva

A atividade clínica dirigida às condições genéticas acompanhou os avanços tecnocientíficos na compreensão dos mecanismos genéticos das doenças e do uso de testes de suscetibilidade genética, bem como o reconhecimento das suas implicações psicossociais para o indivíduo.

Associações profissionais produziram diretrizes a propósito da utilização clínica dos testes genéticos e vários países possuem legislação relativa ao seu enquadramento e uso (Kääriänen *et al*., 2010). Têm sido igualmente produzidas recomendações de organismos governamentais para a uniformização das definições dos testes genéticos nas suas diversas aplicações (Pinto-Basto *et al*., 2010; Sequeiros, 2010). Resulta, assim, que o aconselhamento genético é tido como parte integrante e condição necessária à realização de testes genéticos, percorrendo, com objetivos específicos, as fases pré- e pós-teste (Kääriänen *et al*., 2010).

Torna-se relevante, neste contexto, como refere Zagalo-Cardoso (1988, *in* Rolim, 2007), distinguir doenças genéticas de doenças hereditárias: por doenças hereditárias consideram-se as afeções transmitidas de uma geração à geração seguinte por intermédio do genoma (isto é, de todo o material genético contido nos cromossomas); doenças genéticas designam as afeções "*transmitidas hereditariamente, cuja emergência se deve, exclusivamente, a mutações num ou mais genes (designadas, respetivamente, doenças monogénicas ou poligénicas)*" (Rolim, 2007: 4)[8].

[8] Se as doenças hereditárias, como se depreende, são doenças genéticas, nem todas as doenças genéticas são hereditárias, como em casos de doenças com padrões de transmissão autossómica recessiva. Nestes casos (como fibrose quística ou fenilcetonúria, por exemplo), a doença pode estar presente num progenitor homozigoto mas não ser transmissível aos filhos. Zagalo-Cardoso (1988, *in* Rolim, 2007) considera, assim, que as doenças genéticas abrangem todas as doenças hereditárias, bem como as que são causadas por alterações no genoma não existentes nos antecessores.

É comum uma abordagem diferenciada aos testes genéticos em função do contexto e natureza da sua aplicação. Contudo, para fins associados às doenças genéticas humanas e respetivas questões clínicas, sociais e éticas, a seguinte definição é proposta por Harper (1997: 8) como funcional:

> "*Genetic testing is the analysis of a specific gene, its product or function, or other DNA and chromosome analysis, to detect or exclude an alteration likely to be associated with a genetic disorder*".

É relevante diferenciar os testes genéticos de diagnóstico (incluindo os testes genéticos pré-natais e pré-implantatórios[9]) dos testes genéticos preditivos (Harper, 1997). Os primeiros designam o uso de testes genéticos em indivíduos que, por apresentarem sintomas específicos, estão ou podem estar doentes. Os segundos referem-se aos testes genéticos efetuados em indivíduos saudáveis, assintomáticos, incluindo (Kääriänen *et al*., 2010): i) testes pré-sintomáticos, em que um indivíduo saudável é testado para uma condição monogénica com penetrância[10] completa ou quase total (a doença de Huntington[11] ou cancros hereditários de elevada penetrância, como a polipose adenomatosa familiar [PAF], por exemplo); e ii) testes de suscetibilidade ou de predisposição, quando o risco de ocorrência de uma doença multifatorial (isto é, dependente da interação poligénica e do efeito de estilos de vida e fatores ambientais) é variável, como na maioria dos cancros[12].

Os testes genéticos para cancros hereditários permitem a identificação de mutações ou polimorfismos genéticos associados a maior probabilidade de a doença ocorrer num indivíduo. Os

[9] Os testes de diagnóstico pré-natal designam o processo de avaliação da doença (ou do seu potencial) em fetos. Envole a colheita de ADN do feto, habitualmente entre as 10 e as 20 semanas de gestação. Os testes de diagnóstico pré-implantatório associam-se a técnicas de reprodução assistida (fertilização *in vitro*), e designam o processo de teste genético e posterior seleção de embriões não-portadores de mutações deletérias específicas e a sua subsequente transferência para o útero materno a fim de prosseguir a gestação normal (Genetics Home Reference, 2012).

[10] A probabilidade de anomalias no genótipo (alterações genéticas) conduzirem a alterações no fenótipo (desenvolvimento da doença) denomina-se penetrância. Dito de outro modo, designa a probabilidade em a doença se manifestar. Por exemplo, na PAF, uma forma de cancro colorectal hereditário (*cf.* pág. 29), a penetrância é praticamente completa (100%), indicando com um grau de certeza elevadíssimo que os indivíduos portadores da mutação genética associada (*APC*) desenvolverão a doença no futuro (Nagy, Sweet, & Eng, 2004).

[11] A doença (ou coreia) de Huntington é uma doença neurológica degenerativa de início tardio. Trata-se de uma doença genética de penetrância praticamente completa, com um padrão de transmissão hereditário autossómico dominante, o que significa que um filho tem 50% de probabilidade de herdar o gene mutado do progenitor. O gene responsável pela transmissão hereditária da doença foi descoberto no início dos anos 1990. O seu estudo e as implicações para doentes e portadores, bem como as dinâmicas familiares subjacentes, são tidos como paradigma na literatura sobre a adaptação psicossocial às doenças genéticas e ao risco que se lhes associa (Brouwer-Dudokde, Savenije, Zoeteweij, Maat-Kiewit, & Tibben, 2002).

[12] Como nota Sequeiros (2010), a designação dos testes genéticos usados em cancros inclui forte ambiguidade. Se a maioria dos testes genéticos em cancros são testes de suscetibilidade, o seu uso em indivíduos saudáveis é pré-sintomático por natureza, particularmente em casos de mutações com elevada penetrância; os testes genéticos para mutações de penetrância reduzida (para os genes *BRCA*1/2 no cancro da mama e ovários, por exemplo) possuem um valor preditivo menor, embora não se considerem meramente testes de suscetibilidade.

benefícios práticos que lhe estão associados envolvem: a promoção de comportamentos de monitorização e redução de risco decorrentes da obtenção de informação sobre a suscetibilidade genética (*e.g.* medidas profiláticas, vigilância ou prevenção); os indivíduos identificados como não portadores das mutações genéticas de elevada penetrância ficam "dispensados" da realização de exames de rastreio invasivos (Rolim, 2007); a identificação de uma mutação genética conhecida num indivíduo sintomático pode ser útil, ainda, na sinalização de familiares saudáveis para aconselhamento oncogenético.

A informação preditiva que deriva dos testes de suscetibilidade é relativa, isto é, indica um grau de vulnerabilidade variável para a doença em análise (Clarke, 1997a; Ever-Kiebooms *et al*., 2000). Comparativamente à população geral, a presença de um ou mais genes de predisposição no genoma de um indivíduo torna-o mais vulnerável à disrupção do seu funcionamento biológico normal e consequentemente ao desenvolvimento da doença em questão. Esta evidência, porém, não permite estimativas de risco que determinem com exatidão quando e sob que circunstâncias a expressão génica pode ocorrer. Numerosos genes de predisposição para doenças específicas possuem expressão extremamente variável (polimorfismos) e encontram-se amplamente distribuídos através das populações humanas. A identificação de tais polimorfismos não é causa necessária ou suficiente para despoletar a doença, sendo assumido que outros genes ainda desconhecidos, os seus produtos proteicos, material molecular e/ou fatores ambientais possam igualmente estar implicados[13] (Lock *et al*., 2006).

Vários autores foram produzindo previsões laudatórias quanto à melhoria das condições de saúde associadas à utilização dos testes genéticos. Embora a comunidade científica enfatize a sua aplicação como esmagadoramente benéfica, alguns autores referem amplas limitações. A grande complexidade inerente à interpretação da informação genética e à sua natureza probabilística estão na base das dúvidas quanto às vantagens e desvantagens dos testes de suscetibilidade genética (Trepanier *et al*., 2004). Hubbard e Lewontin (1996, *in* Rolim, 2007) apontam outras fontes de incerteza associadas à informação veiculada pelos testes de suscetibilidade: variações no modo de hereditariedade genética, imprecisões nas informações dadas pelas famílias quanto aos parentescos biológicos e variações no controlo de qualidade e precisão dos procedimentos laboratoriais. Outra limitação aos testes de suscetibilidade genética para doenças multifatoriais recai no tipo de resultado que pode ser obtido (Clarke, 1997a). A estimativa de risco, comummente calculada de

[13] A correspondência entre genótipo e fenótipo envolve extrema complexidade, sobretudo quando em causa estão condições genéticas multifatoriais, como na maioria das doenças comuns. O ADN interage com outros componentes celulares, como o RNA, enzimas, proteínas e outras muléculas-chave, incluindo iões reguladores, num processo dinâmico ao longo do ciclo de vida dos indivíduos. Esta interação é influenciada por condições intra- e extracelulares e pelo macro-ambiente externo ao corpo do organismo. *Stressores*, toxinas e outros fatores, como o envelhecimento, provocam flutuações no ordenamento celular cuja causa ou origem é extremamente difícil de decifrar com exatidão (Plows, 2011).

acordo com modelos estatísticos especíIficos, pode ser apresentada como sendo um risco geral, veiculado através de uma fração, percentagem ou índice de probabilidades; ou como um risco relativo comparado com a média da população geral, podendo ser estabelecida uma "janela temporal" para o início da manifestação da doença. As restrições à validade das estimativas de risco resultam de enviesamentos nas bases de dados a partir das quais são formuladas, nomeadamente quanto à sua composição em termos de frequência da doença e da população abrangida (maioritariamente mulheres norte-americanas cuja história familiar se encontra documentada), tendendo a negligenciar a variabilidade genética entre diferentes grupos étnicos e populações específicas, não obstante o esforço prospetivo assinalável de inúmeros projetos de investigação na construção de uma base de dados ampla e fiável (Clarke, 1997a; Prior, 2006).

A ambiguidade inerente ao valor preditivo dos testes de suscetibilidades (não prediz quem será afetado, quando, ou se será mesmo, deixando margens de incerteza consideráveis sobre o *timing* do aparecimento de sintomas e a extensão do desenvolvimento da doença) acentua preocupações sobre potenciais consequências psicológicas, físicas e sociais, em indivíduos saudáveis nessas circunstâncias. Algumas dessas preocupações decorrem da promoção da medicalização da vida, geneticização das diferenças individuais e provisão de cuidados de saúde, a que se acrescenta: excessiva consciencialização quanto à saúde individual (obscurecendo possíveis soluções coletivas[14]); ansiedade face à possível eclosão de sintomas, estigma social associado ao estatuto de "estar em risco"; e profundas modificações no modo de pensar a saúde, doença, morte, e na visão ontológica da vida (Lippman, 1992; Peterson & Lupton, 1996).

## 2. ACONSELHAMENTO GENÉTICO: PARA ALÉM DO TESTE MOLECULAR

Seguindo as coordenadas propostas por Evans (2006), a análise da designação "aconselhamento genético" fornece as premissas básicas da sua natureza: primeiro, incide nos genes, assumindo uma componente molecular, científica e especializada; depois, o termo "aconselhamento" (*counselling*) sugere o envolvimento de uma conversação, a criação de um espaço psicológico para pensar sobre determinada questão ou realidade. Consequentemente, as suas fundações conceptuais radicam na

[14] Clarke (1997: 105) enfatiza a natureza interativa entre genótipo e ambiente e que os fatores biológicos são mediados socialmente e, com efeito, transformáveis. O autor sublinha que as populações que vivem em contextos de pobreza tendem a ser excluídas do acesso aos cuidados de saúde em geral e dos cuidados genómicos em particular, e refere: "*social and political decisions that ameliorate poverty and that alter lifestyles appropriately may enhance the health of the general population more effectively and more cheaply than individualized susceptibility screening to identify those at increased risk, although such policies may be unpopular with some because they will generate less profit for the biotechnology and pharmaceutical industries*".

investigação científica, medicina (genética) e comunicação humana, em concreto no estabelecimento de uma relação de ajuda profissional.

Ao situar-se na interface entre o teste molecular, o indivíduo e a família, o aconselhamento genético apresenta complexas implicações éticas, psicológicas e sociais, também patentes na definição dos seus objetivos e enfoque processual e profissional ao longo da sua evolução histórica.

### 2.1. Evolução histórica do aconselhamento genético

O conhecimento de que muitas doenças se transmitem entre gerações remonta a tempos imemoriais. Contudo, apenas no dealbar do século XX o termo "genética" foi pela primeira vez empregue, pelo britânico William Bateson, entusiasta das "unidades de hereditariedade" que Gregor Mendel havia desenvolvido décadas antes, para designar a ciência da hereditariedade (Capra, 1996).

A história do aconselhamento genético pode ser organizada em três grandes paradigmas: eugenia, medicina preventiva, e medicina psicológica. Cada paradigma baseia-se em diferentes conjuntos de assunções quanto aos objetivos, princípios e práticas. A emergência de uma orientação psicológica (e psicoterapêutica) do aconselhamento genético resultou, em parte, da progressiva consciencialização das dificuldades em assegurar princípios orientadores por parte dos outros paradigmas e da afirmação da natureza eminentemente psicossocial das doenças genéticas e do processo do aconselhamento genético. Tal mudança para um paradigma psicológico teve importantes repercussões filosóficas e práticas, refletidas em questões-chave no processo de aconselhamento genético, como a educação genética, aspetos comunicacionais, tomada de decisão, apoio psicossocial e formação profissional (Kenen & Smith, 1995).

O primeiro período da prática médica associada à genética humana surgiu durante as primeiras décadas do século XX e decorreu sob os auspícios eugénicos[15]. A sua ação privilegiava a conservação e aumento da qualidade das populações a nível genético, através do encorajamento da reprodução em pessoas cujo genótipo apresentava alegada superioridade e "desencorajando" a reprodução dos geneticamente menos capacitados, evitando assim a perpetuação de património genético inapto face aos moldes sociais preconizados. Mudanças sociais e políticas durante a II Grande Guerra Mundial, uma compreensão mais sofisticada da genética das populações e o repúdio do genocídio Nazi, colocaram como objetivo primordial da prática da genética a separação da eugenia e das políticas coercivas associadas (Resta, 2006).

[15] A eugenia ocorreu com maior evidência nos Estados Unidos da América, Grã-Bretanha e Alemanha envolvendo medidas propagandísticas de educação pública e estratégias de prevenção de doenças, segregação e esterilização e, no caso da Alemanha sob a égide do regime Nazi, políticas de higienização racial que incluíram a eliminação (Thom & Jennings, 1996).

O paradigma médico-preventivo constitui o segundo momento da evolução histórico-conceptual do aconselhamento genético. Entre o final da década de 1940 e o início dos anos 1960, a prevenção das doenças genéticas focou-se na otimização da saúde populacional, assistindo-se a uma atitude pedagógica centrada na provisão de informação médica no âmbito dos cuidados de saúde materno-infantis e do diagnóstico de crianças com anomalias congénitas (Zagalo-Cardoso, 1995). Esta abordagem tinha subjacente o argumento de que a prevenção das doenças genéticas nas sociedades democráticas apenas teria lugar através de medidas voluntárias. Neste período, o termo "aconselhamento genético" surge pela primeira vez, cunhado por Sheldon Reed, em 1947, para designar a nova área de atuação da genética clínica, com o objetivo de: "(…) *provide people with an understanding of the gentic problems in their family*" (Reed, 1955: 12, *in* Resta, 2006).

A transição do aconselhamento genético dos departamentos académicos, onde era ministrado por técnicos com formação em ciências biológicas, para centros médicos, decorreu a par do progressivo reconhecimento das dimensões psicossociais inerentes às várias formas de doença (incluindo as doenças genéticas) (Kenen, 1984; Shontz, 1975, *in* Kessler, 1979b). Os objetivos do aconselhamento genético começaram a incorporar a importância do alívio da sobrecarga psicológica associada à informação genética e a otimização da compreensão por parte do consultando através do recurso ao *counselling* (Evans, 2006; Kessler, 1979b). Ou seja, a evolução ocorre desde a prevenção da doença genética para uma relação médico-paciente centrada na pessoa e focada na comunicação do risco genético. Kessler (1979b: 20) caracteriza assim o paradigma psicológico do aconselhamento genético:

> "(…) *deals with human behaviors such as health and illness, procreation, parenthood, and, sometimes, life and death. It views the problems posed by a genetic disorder as being intimately related to the overall situation of the persons, their ways of solving problems, making decisions, and adapting to life crisis*".

O aconselhamento genético, com efeito, não obstante as suas raízes na biologia, não se limitou a uma prática exclusivamente médica, evoluindo para uma atividade orientada por pressupostos psicológicos (Kessler, 1979b; Resta, 2006; Weil, 2000). Zagalo-Cardoso (1995) refere que a evolução da teoria e prática do aconselhamento genético se processou a partir da unidisciplinaridade (biologia) para uma conjunção multidisciplinar de várias disciplinas científicas, que atualmente sintetizam de forma inter- ou transdisciplinar uma nova metodologia que contempla a complexidade dos seus objetivos e práticas, ilustrando os três "paradigmas dominantes" do seu percurso histórico.

**2.2. Dois objetivos e um princípio orientador, ou a natureza do aconselhamento genético**

O final da década de 1960 reforçou importantes avanços científicos e tecnológicos no domínio da genética, assistindo-se a uma mudança paradigmática na natureza da prática médica e da relação médico-paciente que se traduziu na erosão da autoridade do clínico (Siegler & Osmond, 1974). No aconselhamento genético, tais transformações potenciaram uma relação médico-paciente assente na facilitação do processo decisório e promoção dos direitos, responsabilidade e autonomia pessoal do paciente (Kessler, 2000c). Em 1969, realizou-se o primeiro programa de formação de *genetic counsellors*[16], no *Sarah Lawrence College* (Nova Iorque, Estados Unidos da América [EUA]), inaugurando um período marcado pela definição conceptual, teórica, clínico-prática e profissional do aconselhamento genético. Para o desenvolvimento profissional dos futuros clínicos, o modelo não-diretivo da terapia centrada na pessoa de Carl Rogers foi eleito como base para as competências de entrevista. Ou seja, como "lente" prática para ajudar os consultandos a tomarem as suas melhores decisões a partir de uma perspetiva pessoal sem interferir ou "guiá-los" até uma decisão em particular (por exemplo, realizar ou não um teste genético, ou terminar ou prosseguir uma gravidez) (Weil, 2000).

Seymour Kessler, a partir das contribuições de Szasz e Hollender (1956, *in* Kessler, 1979b) para a filosofia da medicina a propósito dos modelos de relação médico-paciente, destaca o modelo de participação mútua como o mais compatível com o aconselhamento genético não-diretivo, pois opera no sentido da capacitação do consultando. Paralelamente, significativas mudanças políticas e sociais, concretizadas numa acrescida proactividade reivindicativa dos movimentos pró-direitos civis, *gay* e feministas, incluindo de pacientes e portadores de deficiência, propiciaram que os pressupostos não-diretivos se assumissem como componente central da teoria e prática do aconselhamento genético (Weil *et al*., 2006). Neste contexto, o *Ad Hoc Committee on Genetic Counseling*, da *American Society of Human Genetics* (ASHG), estabeleceu, em 1975, a definição de aconselhamento genético considerada até hoje como a mais consensual e influente (Resta, 2006 *in* Rolim, 2007: 204):

> "*O aconselhamento genético é um processo comunicacional que lida com os problemas humanos associados à ocorrência, ou risco de ocorrência, de uma doença genética numa família, envolvendo a tentativa, por parte de uma ou mais pessoas adequadamente treinadas, de ajudar a pessoa ou família a: i) compreender os factos médicos, incluindo o*

[16] A designação "oficial" de *genetic counsellor*, em Português, é, ainda, inexistente. Assiste-se a um consenso informal em torno do uso do termo "profissional de aconselhamento genético", embora outros sejam, contudo, igualmente utilizados: assessor genético (consultor ou aconselhador genético) designa um especialista não-médico que, nos países onde existe, fornece informação e apoio (aconselhamento genético) às pessoas preocupadas com uma doença que pode ter uma base genética (EuroGentest, 2008).

> *diagnóstico, o curso provável da doença e as formas de tratamento disponíveis; ii) tornar claro o modo como a hereditariedade contribui para a ocorrência da doença e o risco de recorrência em familiares; iii) compreender as alternativas para lidar com o risco de recorrência; iv) escolher o plano de ação que lhes parece mais adequado, tendo em conta o seu risco, os seus objetivos familiares e os seus valores éticos e religiosos, agindo de acordo com essa decisão; e v) facilitar a melhor adaptação possível à doença, num familiar afetado, e/ou ao risco de recorrência dessa doença*".

A definição proposta denota a articulação de diversos aspetos centrais do aconselhamento genético, conjugados em dois objetivos nodais que intersetam a conceptualização da prática: por um lado, estabelecer canais comunicacionais efetivos entre clínico e consultando para a adequada educação deste último e capacitação da tomada de decisões informadas; e, por outro, facilitar a adaptação psicossocial do consultando e seu bem-estar psicológico. Embora sem ter sido explicitamente mencionado, a definição proposta suporta as premissas não-diretivas ao aconselhamento genético. Também o Código de Ética da norte-americana *National Society of Genetic Counselors* (NSGC) elege o "*respect of clients' beliefs, cultural traditions, inclinations, circumstances, and feelings* (…) *and enable clients to make informed independent decisions, free of coercion*" (NSGC, 1992: 42, *in* Weil *et al*., 2006: 201) como aspeto central. A não-directividade é entendida como princípio orientador e não como um objetivo, de acordo com o que pode ser depreendido do enfoque na qualidade do processo comunicacional entre clínico e consultando.

Barbara Biesecker (2001), numa revisão sobre a evolução temporal das definições do aconselhamento genético, condensa as suas mais proeminentes premissas conceptuais na proposta de um paradigma psicoeducativo enquanto definição contemporânea da prática. Especificamente, o aconselhamento genético é visto como processo dinâmico, psicoeducativo, centrado na disponibilização de informação, com o propósito de potenciar a capacidade de o paciente a usar de forma personalizada, ajudar a minimizar o *stress* e a incrementar o controlo pessoal. A autora recomenda que os objetivos do aconselhamento genético sejam adaptados às especificidades das suas diferentes subespecialidades (contextos de reprodução humana, pediátrico / adulto e doenças comuns), bem como às necessidades dos consultandos, contemplando as peculiaridades socioculturais e as inerentes aos serviços de saúde de cada país.

Em síntese, as distintas definições propostas realçam três componentes fundamentais do aconselhamento genético entrelaçadas na senda de uma prática apropriada: i) educativo e informativo; ii) apoio psicológico e ajuda na solução de problemas; e iii) facilitar a tomada de decisões informadas. As várias definições de aconselhamento genético refletem uma atividade

complexa. De facto, apesar de ter emergido no contexto da medicina, assumiu-se ao longo do tempo como uma disciplina híbrida, situada na fronteira entre os domínios biomédico e psicossocial. Assim, atua a partir de premissas que assentam na necessidade de personalizar informação altamente científica numa linguagem compreensível, que possa ser facilmente assimilada, intelectual e emocionalmente, por indivíduos e famílias, num percurso translacional do laboratório à família e, de modo mais abrangente, à sociedade.

### 2.3. Debates contemporâneos: redefinindo o aconselhamento genético e a não-directividade

Kessler (2000a) descreveu duas abordagens comuns associadas à prática do aconselhamento genético: um modelo de ensino, alinhado com a vertente mais académica da medicina; e um modelo de aconselhamento, conotado com profissões ligadas à saúde mental. O primeiro postula a educação dos consultandos como o objetivo primordial, pressupondo que procuram aconselhamento genético para se informarem sobre a sua (potencial) doença genética. Providenciar informação científica de modo neutro e racional, para que os consultandos possam tomar as suas decisões, é a tarefa primordial. O âmbito da prática é a tradição biomédica ocidental, em que médico e paciente mantêm uma relação de poder assimétrica, análoga à estabelecida entre professor e aluno e assente num nível de expressão emocional limitado. O modelo de aconselhamento, por seu turno, centra-se na compreensão das complexas motivações do consultando para o aconselhamento genético, procurando aumentar o seu sentido de competência e autonomia. Incorpora os conceitos da abordagem centrada na pessoa de Carl Rogers (1974) para potenciar a adaptação do consultando ao (potencial) risco genético, bem como um sentimento de controlo sobre a sua vida. A provisão de informação é contemplada mas não é o objetivo central. A autoridade do clínico é secundarizada em detrimento da mutualidade e os consultandos são encorajados a assumir um papel proactivo na consulta.

Vários estudos revelam a adoção dos propósitos não-diretivos nas atitudes dos profissionais do aconselhamento genético (Bartels, LeRoy, McCarthy, & Caplan, 1997), embora outros, sobretudo de natureza qualitativa, apontem para desvios na sua aplicação (Michie, Bron, Bobrow, & Marteau, 1997), colocando algumas questões à sua efetiva viabilidade e pertinência. Face a estas limitações, Kessler (2000c) sublinhou a impossibilidade em extrair a não-directividade do contexto Rogeriano e da sua essência psicoterapêutica, pois o profissional, para além de não ser psicoterapeuta (e de, pelo menos, no contexto do aconselhamento genético não estar investido desse papel), não pode delegar toda a "responsabilidade" do curso do processo no paciente. O autor defendeu a necessidade de mais e melhor formação para os profissionais, sobretudo em técnicas e competências de entrevista, pois a não-directividade repousa em procedimentos inerentes ao *counselling*: um processo interativo que envolve mais do que a supressão de determinações

diretivas ("dar conselhos") e fornecer informação clínica, tendo como objetivo a coconstrução de um caminho suscetível de incrementar o controlo sobre as decisões dos consultandos através de um modo personalizado e coerente de pensar essa informação.

Elwyn, Gray, e Clarke (2000), Kessler (2000b; 2000c), e Weil (2000; 2006) estão entre os autores que de modo mais claro sintetizam a necessidade de reexaminar a não-directividade no aconselhamento genético e de avançar face ao que consideram ser os seus contornos mais problemáticos. Os primeiros ajudam a desvelar alguns dos motivos por que clínicos tendem a descrever a sua prática como sendo não-diretiva (Elwyn *et al*., 2000: 135):

> "*There are good reasons why clinical geneticists and genetic counsellors might wish to claim that their work is non-directive. Such a stance is in keeping with the contemporary dominance of autonomy over the other recognised principles of medical ethics. It protects the profession from an easy confusion with, and moral contamination from, the eugenics movement, and this will be useful to genetic counsellors both in public debate and internally within themselves. It protects the counsellors from over-involvement with clients and perhaps also from litigation. The definition of nondirectiveness that we have presented so far, however, is of only limited use. It does not amount to an operational definition that enables us to recognise nondirectiveness in practice, and it does not enable us to tackle the question of what guidance of clients by counsellors might be appropriate.*"

No âmbito do aconselhamento oncogenético destacam-se algumas circunstâncias quanto a esta questão, nomeadamente o facto de existirem tratamentos disponíveis que reduzem significativamente o risco de ocorrência da doença, em contraste com várias doenças genéticas monogénicas, tidas como afeções genéticas "originais" e sobre as quais o aconselhamento genético fez inicialmente recair a sua prática. Considere-se casos em que o clínico recomenda explicitamente um determinado curso de ação ao consultando considerando o seu melhor interesse (para aceitar um tratamento profilático, por exemplo), ou dos seus familiares (a divulgação de informação genética potencialmente relevante para o risco genético de outros familiares ou dos seus filhos). A tomada de decisões no aconselhamento oncogenético envolve questões de extrema complexidade ligadas ao valor dos testes genéticos e de intervenções e práticas médicas com graus de incerteza significativos. Nestas circunstâncias, a experiência e o conhecimento do clínico são fatores decisivos no auxílio da tomada de decisão do consultando. Sugere-se que as necessidades dos consultandos podem ser asseguradas de modo mais efetivo através de uma "tomada de decisão partilhada", um processo interativo no qual o clínico faz recomendações e o consultando retém um papel crucial na decisão final (Elwyn *et al*., 2000).

Weil (2003) refere três limitações centrais à não-directividade: i) ser definida em torno do princípio da neutralidade, que tende a especificar o que o clínico *não deve fazer*; ii) a sua insuficiência ética enquanto *ethos* central do aconselhamento genético, dadas as implicações individuais, familiares e sociais das decisões envolvidas; iii) a evidência da impossibilidade em ser alcançada em absoluto, pois, no limite, será um *esforço direcionado*, tendo em conta a directividade "inadvertida", "institucional" ou a "meta-directividade". O autor argumenta a sua substituição como elemento central da prática e conceptualização profissional do aconselhamento genético, embora mantendo os seus princípios orientadores. Propõe à comunidade científica uma ampla reflexão e discussão sobre essa questão, avançado que "*the central ethos of genetic counseling should be to bring the psychosocial component into every aspect of the work*" (Weil, 2003: 207).

À medida que o debate sobre a conceptualização contemporânea do aconselhamento genético prosseguia, a NSGC, dos EUA, em 2003, reconhecendo a necessidade de uma prática alicerçada em princípios comuns face ao advento da medicina genómica e consequente expansão da sua área de atuação até às doenças "comuns" (como os cancros), incluindo práticas laboratoriais, biotecnologia, saúde pública e investigação psicossocial, redefine o aconselhamento genético (Resta *et al*., 2006: 79):

> "*Genetic counselling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates, i) interpretation of family and medical histories to assess the chance of disease occurrence or recurrence; ii) education about inheritance, testing, management, prevention, resources and research; and iii) counselling to promote informed choices and adaptation to the risk or condition*".

### 2.4. Em busca do aconselhamento genético ideal: da investigação à formação profissional

Os aspetos referidos anteriormente acentuaram a importância da avaliação das práticas do aconselhamento genético e os serviços envolvidos. Wang, Gonzalez, e Merajver (2004) propuseram um conjunto de temáticas que pretendem enformar os critérios avaliativos do aconselhamento genético, iluminando a investigação e direções futuras em relação às variáveis processuais inerentes aos consultandos (autocontrolo percebido, satisfação das expectativas do paciente, tomada de decisões informadas, qualidade da relação clínico-paciente) e variáveis inerentes aos serviços de genética (nível de formação dos vários profissionais envolvidos). Alguns investigadores, associações profissionais e entidades reguladoras têm seguido este trilho, centrando-se: na avaliação dos processos e resultados da prática do aconselhamento genético, na perspetiva dos clínicos e consultandos e recorrendo a diversas metodologias (Gale, Pasalodos-

Sanchez, Kerzin-Storrar, Hall, & MacLeod, 2010; Henneman, Marteau, & Timmermans, 2008; Hernandez, Selber, & Tijerina, 2006; McAllister, 2007; McAllister, Dunn, & Todd, 2010; Michie, Smith, Heaversedge, & Read, 1999); na avaliação dos serviços de genética (EuroGentest, 2010; Farndon & Bennet, 2008; Nippert *et al*., 2011); e na formação de profissionais (Skirton, Lewis, Kent, Coviello, & the members of Eurogentest Unit 6 and ESHG Education Committee, 2010).

Face à proliferação de práticas desadequadas e à descoordenação de serviços prestados, e também à multiplicidade de sistemas de saúde existentes, foi criado um projeto europeu – EuroGentest – envolvendo a participação de especialistas com diversas formações teóricas e práticas, visando incrementar a qualidade do aconselhamento genético disponibilizado através da certificação e acreditação de profissionais e serviços nos países europeus (EuroGentest, 2011). Assim, a harmonização de objetivos e práticas foi antecipada como tendo o potencial para estabelecer uma base comum, com crescente enfoque na formação e educação dos vários profissionais envolvidos. Rantanen e colaboradores (2008) reviram as linhas orientadoras do aconselhamento genético de várias organizações europeias e produziram um conjunto de recomendações para harmonizar a prática. O aconselhamento genético "ideal", com efeito, deverá contemplar (Rantanen *et al*., 2008): i) um profissional adequadamente formado nos aspetos médicos, éticos e psicossociais; ii) que disponibilize informação relevante e objetiva sobre a doença e o processo clínico; iii) assegurando que o consultando compreende; iv) disponibilizando apoio psicológico; v) obtendo consentimento informado; e vi) garantindo a confidencialidade da informação e prevenindo a discriminação potencialmente resultante; vii) considerando as possíveis implicações familiares; e viii) promovendo um processo de tomada de decisões autónomo.

A nível político-governamental, em setembro de 2010, o subcomité para a Saúde da União Europeia (Council of Europe, 2010) produziu recomendações para os estados-membros quanto ao impacto da genética na organização dos serviços de saúde e formação dos profissionais. Este organismo refere que o aconselhamento genético encerra uma prática inerente aos serviços de genética, destacando-se por: i) decisões informadas quanto aos testes genéticos e suas implicações; ii) envolvimento de profissionais de saúde apropriadamente formados; e iii) caráter interativo, detalhado e adaptado às características específicas do consultando, considerando a maximização da compreensão do complexo contexto genético.

Skirton, Voelckel, e Patch (2010), na esfera da Sociedade Europeia de Genética Humana, publicaram recomendações para a atuação dos profissionais do aconselhamento genético: i) identificar necessidades do indivíduo ou família e usar uma abordagem centrada na pessoa; ii) recolher, selecionar e interpretar informação relevante (história médica família, resultados laboratoriais e dados de investigação); iii) ajudar os pacientes na adaptação às implicações médicas, psicológicas, familiares e sociais da doença ou risco genético; iv) avaliar a probabilidade de

ocorrência da doença; v) providenciar informação e recursos educativos a indivíduos e famílias sobre aspetos hereditários, testes genéticos, gestão e prevenção da doença ou risco genético; vi) promover decisões informadas e a adaptação psicológica dos consultandos e/ou famílias; e vii) facilitar o acesso a recursos adequados para a gestão da saúde a indivíduos ou famílias. Para assegurar tais competências práticas e científicas foi desenvolvido um modelo de conteúdos para a formação teórica e prática dos profissionais através do nível mínimo equivalente a Mestrado.

### 2.5. Considerações éticas

Desde a introdução dos primeiros programas de rastreio neo-natal até aos sofisticados meios moleculares de deteção diagnóstica dos dias de hoje, a genética desencadeou inquietações sociais e complexas questões éticas. Embora, como vimos, a tese do excepcionalismo genético tenha granjeado consideráveis críticas, o caráter excecional da informação genética vigora para efeitos ético-legais, sobretudo devido: à sua "extensão familiar", isto é, às eventuais implicações decisivas na gestão da saúde de familiares potencialmente em risco; e aos perigos discriminatórios do acesso a informação genética por terceiros, nomeadamente, seguradoras, entidades empregadoras, empresas de biotecnologia ou o estado (Clarke, 1997b; McEwen, 2006).

De modo análogo a qualquer ação médica, o consentimento informado é fundamental no aconselhemento genético e o procedimento basilar que visa salvaguardar o respeito bioético pelo indivíduo neste contexto (Sequeiros, 2001). O Artigo 12º da Convenção Europeia para a Proteção dos Direitos do Homem e da dignidade do ser humano face às aplicações da biomedicina (Council of Europe, 1997) refere a necessidade de aconselhamento genético apropriado antes da realização de quaisquer testes genéticos. O consentimento informado é suportado ética e juridicamente (Diário da República, 2005; WHO, 2001) e visa assegurar a confidencialidade da informação genética, tal como a autonomia do paciente e a sua primazia na tomada de decisões, incluindo os seguintes aspetos: disponibilização de informação sobre o risco de contrair a doença em causa, incluindo questões relativas à hereditariedade e aos riscos associados aos descendentes e outros familiares; assegurar que os consultandos compreendem os objetivos dos testes genéticos, as alternativas médicas disponíveis, possíveis resultados e seu significado para a minitorização do risco e possíveis implicações psicossociais; e elucidação acerca das limitações associadas à validade da informação genética, evitando o sobredimensionamento das expectativas face aos resultados do teste molecular.

Sequeiros (2001) refere os princípios éticos clássicos e elementares que deverão nortear o processo de aconselhamento genético: autonomia, através do cumprimento do consentimento informado que assegura a consciencialização das opções tomadas; beneficência e não-maleficência, ou seja, um enfoque na melhoria da qualidade de vida, na disponibilização de avaliação e acompanhamento

psicossocial e na definição de critérios de exclusão (para quem os resultados dos testes genéticos possam resultar em dano); veracidade e fidelidade, fundados no modo como a informação é apresentada e discutida, bem como no acompanhamento médico e psicossocial subsequente; e justiça, isto é, a inexistência de barreiras socioeconómicas, etno-culturais ou outras suscetíveis de limitar a equidade na acessibilidade aos cuidados de saúde. A estes princípios bioéticos, erigidos na tradição do Principalismo da ética médica ocidental, associam-se direitos humanos fundamentais: liberdade de escolha, direito à informação completa e exata, direito à privacidade, direito a cuidados médicos de qualidade e direito à não-discriminação, isto é, a garantia de que todas as pessoas, independentemente do seu genótipo, têm o direito de recusar o teste genético, têm o direito de se reproduzirem e de terem acesso ao trabalho, à educação e à solidariedade social adequados.

Se os princípios bioéticos referidos são geralmente consensuais, algumas questões que se prendem com a autonomia e os direitos sobre a informação genética têm sido, porém, amplamente debatidas. No centro do debate figura a conceção utilitarista inerente ao valor relativo da confidencialidade e privacidade da informação genética, que postula a possibilidade de os clínicos a ultrapassarem quando se afigurar importante que o resultado positivo de um teste seja comunicado a alguma(s) pessoa(s) da família do indivíduo analisado para efeitos de gestão da saúde e do risco genético. O dilema reside entre a proteção da confidencialidade dos dados médico-genéticos do consultando e a prevenção de dano nos familiares. Recomendações de associações profissionais internacionais e diretrizes legais, de um modo geral, enfatizam a primazia da manutenção da confidencialidade da informação genética numa perspetiva individualista, embora postulem que esta deva ser quebrada (isto é, sem o consentimento do paciente) perante circunstâncias específicas que se prendem com a gravidade, iminência e a preventabilidade do risco (Knoppers *et al*., 1998). Estas recomendações incluem o esforço do clínico na elucidação e persuasão do consultando (excluindo, naturalmente, a coerção) da importância de os familiares em risco poderem ser informados; por exemplo, a *Human Genetics Commission* (2002), órgão consultivo do governo britânico, sugeriu a promoção da "solidariedade genética" e do altruísmo nesta matéria.

Davey, Newson, e O'Leary (2006), tendo em conta a natureza eminentemente hereditária da informação genética, propuseram o conceito de *familial comity* como uma noção de autonomia relacional, isto é, em que a autonomia individual deve ser considerada a partir da responsabilidade social para com os outros. Boddington e Gregory (2008), por seu turno, enriqueceram o debate em torno da autonomia introduzindo a noção de integridade enquanto forma de obter uma compreensão mais aprofundada sobre o significado ético envolto no processo da comunicação da informação genética. As autoras questionam a proeminência da autonomia sobre a noção de integridade, enfatizando que ambos os princípios figuram na tradição bioética europeia, a par das noções de vulnerabilidade e dignidade. Integridade designa uma noção moral que pressupõe retidão

e consistência moral do indivíduo e inclui uma visão holística do *self* na sua totalidade (*wholeness*), englobando uma articulação coerente entre o mundo interno e as atitudes e ações no mundo externo, ou seja, na relação com os outros. Um outro aspeto diz respeito à contextualização narrativa face à identidade e história de vida do indivíduo, destacando uma perspetiva diacrónica e sincrónica que valoriza a sua multiplicidade de valores no confronto com o processo de transmissão da informação genética.

Na prática profissional, permanecem no entanto dúvidas sobre em que medida e com que meios os profissionais devem encorajar a transmissão de informação genética na família. Este tema tem colocado interrogações sobre quem detém primazia sobre essa informação; dito de outro modo, a quem pertence a informação genética, se ao indivíduo ou à família (Lucassen & Clarke, 2007). O reconhecimento legal mais amplo para os familiares nesta questão advém do caráter presumido de que estes têm o direito de saber, fundado nos princípios éticos da justiça e reciprocidade no processo decisório. Existem no entanto argumentos ao direito a não saber, em que a ignorância sobre a informação genética, se explicitamente ativada, manifesta uma expressão da autonomia individual, designadamente de proteção face a potenciais consequências psicológicas adversas (Adorno, 2003). Também existe evidência de que a pressão para a transmissão da informação na família poderá subverter valores familiares importantes na manutenção do equilíbrio e interdependência relacionais, sobretudo em famílias coesas e "ligadas", cujo funcionamento se funda numa configuração estrutural mais "tradicional" e multigeracional (Juengst, 1999).

A realização de testes genéticos em crianças é outra questão ética particular amplamente debatida. Defende-se que os testes genéticos em crianças apenas deverão ser efetuados quando existem intervenções de natureza médica capazes de beneficiar a criança face a um resultado positivo no teste de suscetibilidade genética (como no caso da PAF, por exemplo) (Clarke, 2010: 19).

> "*(…) when a child is at risk of having inherited a genetic condition from her parents despite appearing healthy, and when there is a clinically effective treatment or a useful programme of surveillance for complications that can be commenced during childhood, than it is clearly, unarguably appropriate for predictive genetic testing to go ahead. It is to the direct benefit of the child for her genetic status to be clarified*".

Na PAF, estas circunstâncias verificam-se pela diminuição considerável do risco de ocorrência da doença e, por outro lado, no contexto de uma família em risco, por permitir distinguir as crianças para quem os procedimentos médicos de vigilância poderão ser benéficos das que podem ser "poupadas" a intervenções invasivas; e ainda, por "aliviar" os familiares da vigilância angustiante face aos possíveis sinais da doença, eventualmente já presentes noutros familiares (Clarke, 1997c).

Não obstante os efeitos adversos que a vivência de cenários de incerteza pode acarretar para os pais e, indiretamente, para as crianças, Clarke e Gaff (2008) defendem que, perante circunstâncias em que não se vislumbram intervenções médicas capazes de beneficiar a criança ou adolescente, a decisão sobre o teste genético deve ser "deixada" para quando a criança tiver idade legal para tal; nessa altura pode inclusive auxiliar na tomada de decisões importantes da sua vida, como encetar uma relação amorosa, plano reprodutivo, planeamento da sua educação ou carreira profissional. Os autores especificam cinco aspetos a ter em consideração: o direito dos pais na requisição do teste genético; a determinação do grau de maturidade da criança e a sua preparação para participar no processo de tomada de decisão; a limitação da capacidade da criança poder realizar uma decisão autónoma no futuro; a perda de confidencialidade quanto ao estatuto genético da criança; e a sua possível estigmatização devido ao resultado do teste.

McKonkie-Rossel e Spiridigliozzi (2004) propõem uma abordagem que compatibilize os princípios Principalistas com o contexto familiar. As autoras sugerem que o processo de tomada de decisão face ao teste genético inclua a exploração aprofundada da opinião da família quanto a esta questão, fundado numa abordagem ética da família nas intervenções médicas.

### 2.6. Organização dos serviços de aconselhamento genético em Portugal

Em Portugal, a lei 12/2005 determina o enquadramento legal sobre informação e intervenção genéticas, bem como das entidades prestadoras de serviços envolvidos na provisão do aconselhamento genético (Diário da República, 2005). A genética médica foi reconhecida pela Ordem dos Médicos como uma Especialidade em 1998 (com internatos iniciados em 2002), dando sequência ao despacho nº 64 do Ministério da Saúde, de 1995. Os progressos dos últimos anos, abrangentes e significativos quanto a ganhos no planeamento e na criação de serviços, não se traduziram, porém, no desenvolvimento dos recursos e da dimensão dos serviços prestados à comunidade. Tal é descrito no documento da Direção Geral de Saúde (2004) que se propôs iniciar a Rede de Referenciação Hospitalar de Genética Médica (RRHGM). Os objetivos visavam contribuir para o desenvolvimento e dotação dos serviços de genética médica com recursos adequados à prestação de cuidados de qualidade a consultandos, doentes e famílias, e melhorar a complementaridade e articulação dos recursos entre instituições.

O Serviço Nacional de Saúde (SNS) contempla, segundo os dados da RRHGM, seis Serviços de Genética Médica que cumprem os requisitos em vigor desde 1995: três no Norte de país (dois no Porto e um em Vila Real), um em Coimbra, e três em Lisboa.

O aconselhamento oncogenético está geralmente envolvido no SNS, sendo disponibilizado em hospitais oncológicos a nível regional, em serviços de genética médica ou de oncologia de hospitais gerais, ou através de institutos públicos. Instituições públicas ligadas ao Ministério da Educação

(como algumas faculdades de medicina), e algumas instituições privadas, providenciam serviço clínico e laboratorial. Existe um Programa Nacional de Teste Pré-Sintomático e Aconselhamento Genético (PNTPSAG), aprovado em 1995, e inicialmente dirigido a doenças neurológicas de início tardio (a Doença de Machado-Joseph foi a primeira doença a ser contemplada no PNTPSAG) servindo de modelo para protocolos de aconselhamento genético noutras doenças, sobretudo ataraxias hereditárias (como a doença de Huntington e a Polineuropatia Amiloidótica Familiar – I, PAF-I) (Sequeiros, 2006). Na fase inicial de implementação do PNTPSAG procedeu-se à formação de equipas dos centros nacionais e elaboração dos protocolos de consultas, que contemplam um conjunto de sessões pré-definidas (pré- e pós-teste pré-sintomático) e incluem avaliação e acompanhamento psicossocial e aconselhamento genético (Sequeiros, 1996).

Em 2010, iniciou-se o primeiro mestrado profissionalizante em aconselhamento genético em Portugal, ministrado pelo Instituto de Ciências Biomédicas Abel Salazar (ICBAS), da Universidade do Porto. Encarando o aconselhamento genético como uma nova disciplina científico-técnica da área da saúde, o objetivo consistiu em formar profissionais para posterior integração em equipas clínicas multidisciplinares de genética médica, de acordo com as premissas essenciais ao desenvolvimento de competências adequadas à prática do aconselhamento genético (Paneque & Sequeiros, 2010; Skirton, Voelckel, & Patch, 2010).

## 3. CANCROS HEREDITÁRIOS E TESTES DE SUSCETIBILIDADE GENÉTICA

Com a identificação dos genes de suscetibilidade para diversos cancros, passaram a estar disponíveis testes genéticos através de técnicas laboratoriais de análise molecular que permitem o diagnóstico da predisposição para essas doenças. As síndromes hereditárias de cancro predispõem os indivíduos a cancros comuns, como cancro da mama, ovários ou intestino, e a tipos de cancro mais raros, como a síndrome de Peutz-Jeghers ou o cancro gástrico difuso.

Os cancros hereditários são um bom exemplo da nova genética e da sua aplicabilidade: doenças com um padrão de transmissão hereditário autossómico dominante[17], poligénicas e multifatoriais. A transmissão autossómica dominante traduz um risco genético de 50% de cada filho herdar, do progenitor afetado, a(s) mutação(ões) genética(s) responsável(eis) pela doença e de a vir a

[17] Embora a esmagadora maioria das sínromes hereditárias ou familiares de cancro possuam um padrão de transmissão hereditário autossómico dominante, existem algumas síndromes, consideradas raras, cuja transmissão hereditária é recessiva, variável, ou incerta (Lindor, McMaster, Lindor, & Greene, 2008).

desenvolver, com maior probabilidade, ao longo da vida (Harper, 1998)[18]. A etiologia das doenças poligénicas, isto é, em que estão envolvidos vários genes, é multifatorial, pois depende da interação de múltiplos genes e do efeito de estilos de vida e fatores ambientais. Aos cancros hereditários associam-se, assim, níveis de risco ou de predisposição genética variáveis que condicionam as potenciais consequências médicas e as medidas profiláticas disponíveis (Patenaude, 2005). Schneider (2002) enfatiza a redução de custos da identificação de indivíduos em risco, pois os não portadores não incorrem numa suscetibilidade aumentada, não sendo necessário empregar meios de rastreio, mas somente medidas de vigilância sugeridas à população em geral.

Os riscos de desenvolver a doença associados às síndromes hereditárias de cancro podem ser consideravelmente mais significativos do que os da população geral, para além de que os riscos de múltiplos cancros e de a doença ocorrer cedo são igualmente elevados. A maioria dos cancros são esporádicos, isto é, desenvolvem-se sem risco familiar ou hereditário (Schneider, 2002). Os cancros hereditários são aqueles em que mutações genéticas já identificadas como estando implicadas no desenvolvimento da doença são transmitidas hereditariamente. Constituem um fator de risco primário e correspondem a 5 a 15% da totalidade dos casos de cancro diagnosticados. Nas famílias com presença da forma mutada do gene responsável pela suscetibilidade, a percentagem de pessoas afetadas pode chegar aos 50% (Schneider, 2002). Os cancros hereditários, para além dos aspetos especificamente clínicos, caracterizam-se por: precocidade do aparecimento (antes dos 50 anos de idade); ocorrência do mesmo tipo de cancro em vários membros da família ou de mais do que um tipo de cancro na mesma pessoa; elevado número de casos numa família; e proximidade do parentesco entre os indivíduos afetados (Regateiro, Silva, & Lemos, 2002; Sifri, Gangadharappa, & Acheson, 2004).

### 3.1. Perfil biomédico dos cancros hereditários

Esta tese integra estudos com indivíduos e famílias portadores de mutações genéticas associadas às formas hereditárias de cancro mais frequentes: cancros da mama e ovários e cancro colorectal[19]. Apresentam-se de seguida os aspetos mais relevantes para a contextualização biomédica das doenças em apreço nesta tese.

[18] A transmissão autossómica recessiva significa que para uma pessoa poder vir a desenvolver a doença tem que herdar duas cópias alteradas do mesmo gene, uma de cada progenitor. Em casos de transmissão de uma cópia normal e de uma cópia alterada do gene, o sujeito será portador saudável, na maior pare dos casos, sem a doença se manifestar. São exemplos de doenças autossómicas recessivas a fenilcetonúria ou a fibrose quística (Harper, 1998).

[19] Refira-se que o estudo *Experiencing genetic counselling for hereditary cancers: the client's perspetive* (*cf*. capítulo I, pág. 69) inclui pessoas que aguardavam o resultado do teste de suscetibilidade, desconhecendo, como tal, o seu estatuto genético aquando da participação no estudo. Por outro lado, o estudo intitulado *Families' experience of oncogenetic counselling: accounts from a heterogeneous hereditary cancer risk population* (*cf*. capítulo I, pág. 83) contou com a participação de uma família em que alguns elementos eram portadores de mutações de suscetibilidade a cancro gástrico difuso hereditário.

### *3.1.1. Cancro da mama e ovários*

O risco cumulativo de uma mulher desenvolver cancro da mama é de cerca de 10%, nos países industrializados; entre aproximadamente 5 a 10% dos cancros da mama devem-se à transmissão hereditária de genes de suscetibilidade (Hodgson *et al*., 2007). Os genes de suscetibilidade que surgem com maior incidência no cancro da mama e/ou ovários são denominados *BRCA*1[20] e *BRCA*2 e encontram-se nos *loci* dos cromossomas 17q21 e 13q12, respetivamente.

A ocorrência de mutações do gene *BRCA1* é responsável por cerca de 50% dos casos de cancro da mama e cerca de 2% dos casos de cancro em mulheres[21]; mutações em *BRCA2* aumentam igualmente a suscetibilidade para o cancro da mama (Regateiro *et al*., 2002). A penetrância destas duas mutações envolve incertezas; estima-se que o risco cumulativo de indivíduos portadores de uma destas mutações pertencentes a famílias com elevado risco é de: 60 a 85% para o cancro da mama; 20 a 60% para cancro do ovário para as mutações em *BRCA1*; e 10 a 27% em *BRCA2* (Antoniou *et al*., 2003). Para além disso, no incício do século XXI existiam mais de 300 mutações identificadas em *BRCA*1, sem que todas tivessem correspondência a consequências deletérias (Prior, 2006). Mutações genéticas associadas ao gene *BRCA2* predispõem o portador a um espectro alargado de outros cancros, incluindo próstata, estômago, tubo falopiano, pâncreas e tumores na mama masculina (Hodgson *et al*., 2007). Outros genes críticos para formas hereditárias de cancro da mama são o gene *MYC*, associado a cerca de 20% dos casos, *TP53* (25-36%) e *ERBB2* (30%) (Hodgson *et al*., 2007). É de referir a suscetibilidade acrescida para cancro da mama e ovários decorrente de outras síndromes oncológicas hereditárias, como: Ataxia Telangiectasia, síndrome de Cowden, síndrome de Li-Fraumeni, síndrome de Peutz-Jeghers e cancro do cólon não polipódico hereditário (Regateiro *et al*., 2002; Sifri *et al*., 2004).

As medidas preventivas e de deteção precoce para portadores são recomendadas mais cedo e com maior regularidade do que para a população geral e incluem o autoexame regular, mamografia, ecografia concomitante (vigilância para o cancro da mama), e avaliação pélvica e ecografia transvaginal (vigilância para o cancro do ovário) (Regateiro *et al*., 2002; Schneider, 2002). Existem também intervenções profiláticas que reduzem significativamente o risco de ocorrência da doença: quimioprevenção (Tamoxifeno, Raloxifeno) (cancro da mama), contracetivos orais (cancro do ovário) e cirurgias, casos da mastectomia total bilateral (com ou sem reconstrução mamária), que pode reduzir até 90% do risco de cancro da mama, e da salpingo-ovorectomia bilateral, com um potencial de redução do risco de cancro do ovário até 85% (Black & Smith, 2005-2006, *in* Werner-Lin, 2007). As medidas profiláticas descritas, apesar da eficácia na diminuição do risco de cancro

[20] BRCA é abreviatura do inglês *breast* cancer (cancro da mama).

[21] Existem alguns casos reportados, de valor residual, de mutações do gene *BRCA*1/2 em homens, que predispõe os portadores, igualmente, ao cancro da próstata e cólon. A monitorização médica recomendada prevê a examinação clínica anual da mama, bem como o exame estandardizado da próstata.

da mama e ovários e da mortalidade associada, são soluções "imperfeitas" para a preservação da saúde: a ovorectomia, por exemplo, induz a menopausa cirúrgica e aumenta o risco de osteoporose e doença cardíaca (Kauff *et al*., 2008, *in* Werner-Lin, 2010). Adicionalmente, comportam níveis de tensão psicológica significativos: não eliminam totalmente os riscos de desenvolvimento da doença; são irreversíveis; comprometem, no caso da remoção dos ovários, o plano reprodutivo, e a mastectomia tem implicações adversas na sexualidade, autoconceito e satisfação com a imagem corporal (Howard, Bottorff, Balneaves, & Kim-Sing, 2010). Tais intervenções cirúrgicas são efetuadas num contexto de risco familiar elevado e propostas a partir dos 35 anos de idade ou 5 anos mais cedo do que a idade em que se verificou o caso mais precoce de cancro na família (Regateiro *et al*., 2002). O significado simbólico associado à supressão de uma parte saudável do corpo mantendo a "causa" da doença (gene) é também referido (Stiefel *et al*., 1997, *in* Rolim, 2007).

### *3.1.2. Cancro colorectal*

Entre 10 a 20% dos cancros colorectais são hereditários ou familiares (Helm *et al*., 2003). As síndromes mais comuns deste tipo de cancros são o cancro colorectal não polipódico hereditário (CCNPH) (ou síndrome de Lynch tipo II) e a polipose adenomatosa familiar (PAF). O primeiro corresponde de 2 a 3% dos casos e o segundo a cerca de 1%. Ambas estas formas hereditárias de cancro são caracterizadas pelo desenvolvimento de múltiplos tumores primários numa idade precoce (Meropol *et al*., 2006). Os genes *MLH1* e *MSH2* estão implicados na predisposição ao CCNPH em cerca de 90% das famílias com forte historial médico associado; o gene *APC* está associado à PAF (Scott, 2003).

Indivíduos portadores de mutações implicadas no CCNPH têm um risco estimado entre 34 e 70% de desenvolverem a doença até aos 70 anos e risco adicional para outros tumores associados, tais como no endométrio, estômago, trato biliar, ovários e trato urinário (Helm *et al*., 2003). Os indivíduos em risco e portadores assintomáticos são aconselhados a realizar colonoscopias bi-anuais entre os 20 e os 25 anos, considerando, para os últimos, a vigilância das zonas que potencialmente poderão também desenvolver tumores: endométrio (histeroscopia com biopsia dirigida anual), ovários (ecografia transvaginal anual), estômago (endoscopia digestiva alta), trato urinário (citologia, ecografia e citoscopia) e trato biliar (ecografia) (Helm *et al*., 2003; Regateiro *et al*., 2002).

No caso da PAF, o desenvolvimento de múltiplos pólipos adenomatosos na mucosa intestinal pode ocorrer a partir dos 10/12 anos de idade; no caso de surgirem pólipos, e sem o recurso a intervenções profiláticas, o risco da doença se manifestar até aos 40 anos é de praticamente 100%

(Meropol *et al*., 2006). Portadores da mutação de suscetibilidade têm um risco de 50 a 90% de desenvolver adenomas dudodenais e entre 10 a 15% de risco de ocorrência de tumores desmóides (Scott, 2003). Aos indivíduos em risco e portadores assintomáticos é recomendada vigilância endoscópica; a cirurgia profilática é usualmente proposta entre os 15 e os 25 anos, implicando a remoção dos pólipos, ou, no caso de serem muito numerosos, a colectomia (Helm *et al*., 2003).

#### *3.1.3. Outras síndromes oncológicas hereditárias*

Existem mais de 40 síndromes hereditárias de cancro descritas (Schneider, 2002). Cancro gástrico difuso, mola-melanoma múltiplo familiar atípico (MMMFA), paraganglioma familiar ou cancro hereditário da próstata, são exemplos de síndromes oncológicas que apresentam características semelhantes à maioria dos cancros da mama e ovários e colorectal quanto à idade de aparecimento (sobretudo na idade adulta), penetrância (incompleta) e existência de meios de diagnóstico e rastreio médico. Atualmente, não existem testes de suscetibilidade para o cancro da próstata; no caso de MMMFA o teste disponível possui escasso valor informativo. Outras síndromes cuja idade de aparecimento antecede a idade adulta, tal como na PAF, embora com diferenças quanto à existência de meios de rastreio efetivos, são os casos das neoplasias endócrinas múltiplas (de tipos I, IIa e IIb), a doença de Von Hippel Landau e as síndromes de Peutz-Jeghers ou Li-Fraumeni (Schneider, 2002).

### 3.2. Aconselhamento oncogenético: as partes e o todo

O aconselhamento oncogenético designa o processo de identificação e aconselhamento (*counselling*) a indivíduos com risco acrescido de desenvolver cancro, distinguindo entre os que possuem risco elevado, dos que possuem risco moderado e risco semelhante ao da população geral (Riley *et al*., 2012). Através do recurso à análise da história médica familiar (ou *pedigree*), testes genéticos, testes bioquímicos e imagiológicos, síndromes hereditárias de cancro são identificadas e os riscos de desenvolver a doença são quantificados para o indivíduo em questão (consultando) e seus familiares. A informação gerada visa a criação um plano clínico de gestão do risco, podendo envolver vigilância médica e rastreio clínico, prevenção, medidas de redução do risco e notificação dos familiares em risco. A elucidação sobre os aspetos médicos e hereditários da suscetibilidade genética e assistência psicossocial a indivíduos e suas famílias são outros aspetos também contemplados (Trepanier *et al*., 2004).

A suscetibilidade genética para cancros hereditários caracteriza-se pela identificação de uma alteração genética conhecida, numa família, através de estudo molecular, sendo recomendados meios de prevenção, vigilância e diagnóstico precoce aos consultandos. Os indivíduos cuja história

médica familiar é sugestiva de predisposição acrescida a contrair a doença são encaminhados para consultas de aconselhamento oncogenético, segundo critérios específicos. Indivíduos sintomáticos ou em quem a doença já se manifestou, podem também beneficiar de aconselhamento genético tendo com vista à redução do risco de desenvolver um cancro secundário.

O aconselhamento oncogenético pode contemplar vários temas e um número variável de sessões, dependendo dos aspetos clínicos envolvidos, do serviço médico, pedido, do indivíduo e sua família e do profissional. Pode envolver a questão de saber antecipadamente, embora com graus variáveis de certeza, o seu futuro potencial em termos da doença; a confirmação de um substrato genético num diagnóstico previamente estabelecido e respetiva elucidação sobre os seus significados e potenciais consequências para o indivíduo e familiares; ou o aconselhamento genético pré-natal, envolvendo uma história prévia de doença num progenitor (ou em ambos), onde se discute e clarifica o contexto da gravidez em termos de risco potencial para a descendência. A premissa de base, no caso dos cancros hereditários, é a de que existem vantagens na deteção da presença de mutações genéticas numa fase pré-doença (pré-sintomática), pois permite implementar um programa de monitorização para identificar sinais precoces da doença e despoletar tratamentos preventivos (Schneider, 2002).

A estrutura típica do aconselhamento oncogenético envolve (Sifri *et al*., 2004; Trepanier *et al*., 2004): i) recolha da história médica individual e familiar; ii) avaliação psicossocial; iii) avaliação e determinação do risco genético, incluindo medidas de deteção precoce e estratégias profiláticas; iv) teste molecular para síndromes oncológicas hereditárias; e v) recomendações de *follow-up*. Neste processo estão habitualmente envolvidos médicos geneticistas, enfermeiros, profissionais do aconselhamento genético e, nalguns casos, técnicos de saúde mental e de serviço social e de outras especialidades médicas; o trabalho multidisciplinar, ainda que frequentemente sublinhado, nem sempre se verifica na realidade.

A literatura de cariz psicossocial enfatiza os seguintes aspetos no processo aconselhamento genético (Evans, 2006; Kessler, 1979a): existência de uma fase inicial de contacto (*intake phase*) para explorar a natureza do pedido e os motivos da consulta; promoção da compreensão alargada do significado da doença (e do risco) no indivíduo e na família, incluindo crenças individuais e intergeracionais; provisão de informação sobre o padrão de hereditariedade envolvido na doença; e discussão da gestão da ansiedade e características individuais de personalidade e adaptação, incluindo rede de suporte pessoal e consciencialização sobre possíveis consequências acopladas ao conhecimento da informação genética individual. Para além destes aspetos, Zagalo-Cardoso formulou considerações acerca do papel dos psicólogos no contexto do aconselhamento genético, incluindo as seguintes tarefas: i) acolhimento dos consultandos; ii) avaliação psicológica para

auxiliar a abordagem clínica do consultando e que permita avaliar o impacto e significado do risco ou doença genética; iii) assessoria psicológica e psicoterapêutica, facilitadora da função informativa, ajustativa e de apoio em momentos críticos do processo, incluindo seguimento clínico; iv) intervenção consultiva na equipa multidisciplinar, com o objetivo de melhorar o ajustamento psicológico dos elementos da equipa (Zagalo-Cardoso, 1989).

### *3.2.1. Avaliação e diagnóstico da suscetibilidade oncogenética*

Recentemente, a *National Society of Genetic Counselors* (NSGC), dos E.U.A., atualizou as suas recomendações em relação ao aconselhamento oncogenético, incluindo as condições para a realização de testes genéticos (Riley *et al.*, 2012): sugestionabilidade da história médica pessoal ou familiar; a possibilidade de adequada interpretação do seu resultado; a influência na gestão médica do risco oncológico do indivíduo ou outros familiares; e o consentimento informado.

Existem critérios de elegibilidade para a realização do teste de suscetibilidade genética para cancros hereditários, assentes em características pessoais e familiares indiciadoras de síndromes hereditárias de cancro. Em 1991 foram introduzidos os critérios de Amesterdão, para auxiliar na uniformização do recrutamento de sujeitos para diagnóstico clínico oncológico e seleção das famílias para rastreio genético e vigilância médica intensiva; esses critérios foram mais tarde modificados e expandidos para aumentar a acuidade na identificação de potenciais portadores de mutações genéticas (Rolim, 2007).

Para a pesquisa genética de mutações predisponentes de síndromes hereditárias de cancro da mama/ovários, em *BRCA*1/2, Regateiro e colaboradores (2002), com base na IV Reunião de consenso nacional do cancro da mama da Sociedade Portuguesa de Senologia, apontam como *critérios pessoais* (1): i) diagnóstico de cancro da mama em mulheres com idade inferior a 35 anos; ii) especificidades clínicas, genéticas e histológicas do diagnóstico; iii) mulheres com cancro da mama e dos ovários, em que um é diagnosticado antes dos 50 anos; iv) homens com cancro da mama em qualquer idade; ou v) mulheres assintomáticas com idade superior a 25 anos e, pelo menos, um dos seguintes *critérios familiares* (2): a) familiar com cancro da mama e dos ovários, um dos quais diagnosticado antes dos 60 anos; b) familiar com cancro da mama bilateral; c) dois ou mais familiares em primeiro grau com cancro da mama e dos ovários, independentemente da idade de apresentação da neoplasia; d) dois familiares em primeiro grau com cancro da mama ou dos ovários, um deles diagnosticado antes dos 45 anos; e) dois familiares em primeiro grau com cancro dos ovários, e) dois familiares em primeiro grau, um com cancro da mama e outro com cancro dos ovários, na pré-menopausa; f) homem com cancro da mama, independentemente da idade; g) qualquer familiar com mutação em *BRCA*1/2.

Quanto à pesquisa de mutações associadas a síndromes hereditárias de cancro colorectal, de acordo com os critérios de Amesterdão revistos, o sujeito deverá apresentar todos os seguintes critérios (Sifri *et al*., 2004): i) três ou mais familiares com um dos cancros do espectro do CCNPH; ii) um doente deve ser familiar em primeiro grau dos outros dois afetados; iii) duas gerações consecutivamente afetadas; iv) um caso de cancro, pelo menos, diagnosticado antes dos 50 anos; e v) exclusão da PAF. São ainda conhecidos os critérios revistos de Bathesda (Sifri *et al*., 2004), em que o indivíduo deverá preencher os critérios de Amesterdão, com especificações para a idade de aparecimento da doença (40 e 45 anos), para além de características clínicas, genéticas e histológicas específicas do diagnóstico. Para a PAF, existem dois critérios: existência de mais de 100 pólipos adenomatosos colorectais, e menos de 100 pólipos adenomatosos colorectais e um familiar diagnosticado com PAF (Rolim, 2007).

Portadores de genes alterados incorrem num risco aumentado para desenvolver um determinado tipo de cancro ao longo da sua vida, comparativamente com as pessoas que na sua família não são portadoras desse gene. Os elementos não-portadores têm o mesmo risco que a população geral, não significando, todavia, que desenvolvam necessariamente cancro (penetrância incompleta), do mesmo modo que não elimina o risco de patologia oncológica no futuro. (Regateiro *et al*., 2002). Os resultados negativos possuem significados distintos: um resultado negativo para uma mutação desconhecida na família indica resulados inconclusivos que requerem uma interpretação cuidadosa; o resultado negativo para uma mutação já identificada em familiares em primeiro grau indica um "verdadeiro negativo", significando que o indivíduo não possui geralmente um risco acrescido; ou o resultado negativo traduz alterações no ADN do gene que não indicam, no entanto, a sua influência na função génica normal ("variante de significado incerto") (Riley *et al*., 2012).

A transmissão dos resultados dos testes genéticos deve incluir a interpretação personalizada dos resultados, a reavaliação dos riscos de manifestação da doença e a identificação de familiares em risco, independentemente do tipo de resultados obtido (positivo, negativo ou inconclusivo). Dada a complexidade inerente à interpretação dos resultados e às respostas emocionais que podem estar associadas, é recomendado que a transmissão dos mesmos seja realizada pessoalmente e de acordo com as seguintes diretrizes (Riley *et al*., 2012): exploração de dúvidas e preocupações prévias ao conhecimento dos resultados; interpretação dos resultados, salientando as limitações e sensitividade específica inerente ao teste genético efetuado; avaliação das reações do consultando e o seu grau de compreensão da informação veiculada, disponibilizando apoio emocional; exploração da perceção do impacto da informação nos familiares; disponibilizar informação sobre as recomendações para a gestão médica do risco; encaminhamento para outros profissionais de saúde; identificação dos familiares em risco e disponibilização de apoio específico à transmissão de informação genética na

família e de material informativo (cartas ou referência de sítios na *Internet*). Caso o consultando se recuse a partilhar a informação genética com os familiares em risco, recomenda-se, igualmente, que o clínico avalie o potencial ético-legal para exercer o seu "dever de avisar" (*duty to warn*) os familiares em questão.

## 4. DOENÇAS GENÉTICAS E FAMÍLIA: UMA LENTE SISTÉMICA

A Teoria dos Sistemas Familiares (Fuster & Ochoa, 2000; White & Klein, 2002) constitui uma abordagem útil na compreensão da interface entre família e doenças genéticas (McDaniel, 2005; Miller, McDaniel, Rolland, & Feetham, 2006; Peters, Djurdinovic, & Baker, 1999; Peterson, 2005; Rolland & Williams, 2005; Street, Soldan, & Gray, 2000). A complexidade intrasistémica da família envolve dimensões individuais, relacionais, comunicacionais, estruturais, organizacionais e simbólicas, atualizadas recursivamente com sistemas socioculturais numa lógica de funcionamento enquadrada por eixos sincrónicos e diacrónicos. O pensamento sistémico epitomizou um modo de olhar o mundo fundado na compreensão complexa do sistema natural, no modo como sustém e altera o seu equilíbrio, na interdependência das partes que o constituem e no efeito recursivo associado às alterações nas partes (Bateson, 2000; Durand, 1992). Assim, o pensamento sistémico aplicado *aos* genes e ao aconselhamento genético inclui as especificidades recursivas do espectro biopsicossocial: gene – indivíduo – família – sistema de saúde – contexto sociocultural (Engel, 1977).

Desde a década de 1960 assistimos a mudanças sociológicas que vêm sendo cimentadas no sentido do esboroamento da chamada família nuclear tradicional (Relvas & Alarcão, 2002). A variabilidade e fluidez estrutural que caracterizam a família pós-moderna nas últimas décadas do século XX acentuam a dimensão da escolha sobre a biologia. A crescente influência de meios biotecnológicos na prática médica recombinou ligações familiares nem sempre baseadas em relações biogenéticas[22] (Cussins, 1998; Richards, 1996). Por outro lado, a progressiva medicalização da família e do parentesco coloca desafios adicionais aos profissionais envolvidos no aconselhamento genético, intersetando cultura, família e genómica.

De um ponto de vista médico, relações familiares e de parentesco são enquadradas biologicamente em termos da hereditariedade consanguínea, independentemente dos laços relacionais, emocionais ou sociais. Encontra-se bem documentado na pesquisa antropológica o caráter dinâmico e

[22] O recurso à *fertilização in vitro* de embriões, a bancos de gâmetas e a outros tipos de *biobanks*, são exemplos de possibilidades tecnocientíficas que a biomedicina disponibiliza e que reconfiguram as relações familiares biogenéticas.

variabilidade cultural da definição de família, bem como a influência cultural nas crenças em relação à hereditariedade: para muitos, prevalece o primado relacional que privilegia as ligações não-biológicas e não apenas a transmissão hereditária dos genes de geração em geração; a crença de que as semelhanças com características específicas (físicas, de personalidade ou disposicionais) de "um lado" da família ou do indivíduo doente aumentam o risco em contrair a doença é igualmente destacada (Finkler, Skrzynia, & Evans, 2003; Richards, 1996). Uma vez que a exploração da história familiar é um aspeto central no processo de aconselhamento genético, afigura-se importante antecipar potenciais dificuldades em função das incoerências nos significados partilhados entre clínico e consultando em torno de "quem é a família" (McGrath & Edwards, 2009), sendo necessário considerar os múltiplos significados associados à definição "da família", quer os fundados em premissas biológicas, quer sociais.

### 4.1. Centralidade da família no aconselhamento genético: história familiar e transmissão da informação genética

Historicamente, os elementos mais próximos do paciente estão envolvidos no tratamento e gestão da doença, desempenhando papéis e funções de cariz emocional, instrumental e social (Góngora, 2004). Em genética médica, o paciente central é *a família*, cuja ação proactiva pode tornar-se um importante recurso no diagnóstico (Finkler *et al*., 2003). Assiste-se à crescente incorporação da família no processo médico, motivando o enfoque da investigação nos processos familiares de adaptação ao risco genético e no desenvolvimento de modelos de cuidados de saúde centrados na família (McDaniel, Campbell, Hepworth, & Lorenz, 2005; Van Ripper & Gallo, 2005).

A centralidade da família na abordagem clínica às doenças genéticas manifesta-se, desde logo, na indeclinável evidência biológica da rede hereditária consanguínea. Este pressuposto traduz-se na prática do aconselhamento genético através da mais básica técnica usada em genética clínica: a análise da história médica familiar (ou *pedigree*). A sua proeminência para o aconselhamento genético assume diversas facetas: instrumental, porque envolve o desenho da "árvore genealógica" da família, compreendendo usualmente três gerações, num processo análogo ao genograma familiar, classicamente utilizado em terapia familiar para organizar informação biológica, psicológica e interpessoal da família (Bennett, 1999; McGoldrick & Gerson, 1987); processual, porque é parte integrante da dinâmica das consultas, na avaliação do risco e da consequente elegibilidade para a realização de testes genéticos; e relacional, pois é uma tarefa que tende a envolver mais do que um elemento da família (os consultandos podem não conhecer ou recordar toda a informação, necessitando de contactar outros familiares para recolher informações adicionais) (Boeninck, 2008; Richards, 1996).

Vários investigadores no contexto da genética psicossocial sugeriram adaptações do uso do genograma no aconselhamento genético para auxiliar os consultandos sobre que membros da família poderiam beneficiar da informação genética na gestão da saúde (Daly *et al*., 1999; Eunupu, 1997). O *Colored Eco-Genetic Relationship Map* (Kenen & Peters, 2001; Peters, Hoskins, Prindiville, Kenen, & Greene, 2006; Peters *et al*., 2011) revela-se útil para clínicos: ao anotar a informação psicossocial através de cores e símbolos (ao invés das notações relacionais típicas do genograma), identifica relações que providenciam ajuda emocional e instrumental e promove a compreensão da dinâmica familiar, reconhecendo os familiares e outros significativos que desempenham papéis e funções relevantes na gestão relacional e instrumental do aconselhamento genético, e, em particular, da informação genética. Com uma orientação mais próxima da terapia familiar, Penn (1983) propôs a realização do genograma *orientado para a doença crónica* como modalidade de exploração dos aspetos pragmáticos, estruturais, organizacionais e desenvolvimentais da doença na família.

A dinâmica de exploração da história familiar evoca muitas vezes memórias dolorosas de sofrimento e perda. Envolve ainda, frequentemente, a necessidade de o consultando equacionar o contacto com elementos da família cujos laços podem ter enfraquecido ao longo do tempo, com quem se perdeu o contacto ou cuja relação é pautada pelo conflito ou afastamento. Assim, a prática clínica do aconselhamento genético inclui o contexto psicossocial da história da família e o seu legado relacional, emocional e simbólico, em que a informação assume um caráter simultaneamente individual e intergeracional e o risco genético sustém a sua identidade enquanto "assunto familiar" (Novas & Rose, 2000: 490):

> "*Genetic identity is revealed and established only within a web o genetic connectedness, which is overlaid upon a web of family bonds and family memories, with their burden of mutual obligations and caring commitments, and with all the ethical dilemmas they entail. In becoming part of a genetic network, the subject genetically at risk may rethink their relation to their current family – lovers, potential and atual spouses, children, grandchildren and so forth – in terms of risk and inheritance*".

É na complexa rede intrafamiliar que a informação é ou não disseminada (Gaaf & Bylund, 2010). Tipicamente, recai sobre o consultando a responsabilidade de informar os elementos da família sobre o potencial risco genético em que se encontram, sendo este papel encorajado por clínicos e pelo sentimento de auto-responsabilização do consultando em informar os familiares (Claes *et al*., 2003; Keenan *et al*., 2005; Wilson *et al*., 2004). A forma como o processo de comunicação intrafamiliar se desenrola depende da vulnerabilidade e recetividade do consultando e membros da

família, das estratégias e estilos comunicacionais adotados e do impacto da informação genética nas relações familiares. Trata-se de um processo comunicacional complexo, deliberativo e estratégico, que envolve a perceção interindividual formada sobre os fatores descritos, bem como a capacidade de explicação da informação genética e do modo como é assimilada e entendida, que é enquadrado por regras e padrões familiares de funcionamento e pela ponderação do "momento certo" para a trasmissão da informação genética (Gaff *et al*., 2007; Roshanai, Lampic, Rosenquist, & Nordin, 2010). Neste contexto, é relevante perceber que as estruturas tradicionais de família e parentesco ocidentais vigoram: as mulheres são tradicionalmente as "guardiãs" e "transmissoras" da informação familiar, responsáveis pela saúde da família e manutenção dos laços (Richards, 1996), assumindo a responsabilidade pela informação e revelação do risco genético (D'Agincourt-Canning, 2001).

A comunicação intrafamiliar, para além de poder revestir-se de funções de suporte social e de reforço dos laços familiares (Hughes *et al*., 2002), pode também originar segredos (Sobel & Cowan, 2000). A literatura de terapia familiar sistémica salienta o potencial de ameaça do segredo ao desenvolvimento do sistema familiar, nomeadamente através do bloqueio da comunicação clara e direta por via de fenómenos como a triangulação ou coligações (Ausloos, 1996; Imber-Black, 1994). A incerteza inerente às doenças genéticas pode traduzir-se ainda, do ponto de vista da comunicação intrafamiliar sobre o risco, em confusão sobre a privacidade da informação genética e na gestão de opiniões e posições antagónicas entre diferentes elementos da família face à realização de testes genéticos ou sobre o valor da informação genética na gestão da saúde, podendo redundar, com efeito, em dificuldades na gestão da mesma (Bell & Bennett, 2001). A partilha da informação genética pode envolver dilemas ético-morais em relação aos valores da família que se situam entre a responsabilidade de transmissão de informação potencialmente útil à gestão da saúde dos familiares e o respeito pela autonomia individual dos seus membros, nomeadamente (Hallowell *et al*., 2003; Rhodes, 1998): saber ou não saber, o direito a não saber, o dever de cuidar informando ou o receio de alarmar. Esta "agência moral" pode revestir-se de discursos de esperança, de comunicação triangulada ou de preservação da informação até emergirem circunstâncias consideradas favoráveis para "fazer o que deve ser feito" (Hallowell *et al*., 2003).

### 4.2. Do teste molecular à família, passando pelo indivíduo: considerações psicossociais

Existe um reconhecimento crescente entre clínicos e investigadores de que a família influencia e é influenciada pelo sentido atribuído, pela resposta e uso da informação durante a experiência do aconselhamento genético (Doukas, 2003; McDaniel, 2005; Wexler, 1979). O indivíduo potencialmente em risco raramente tem exclusividade na decisão do seu envolvimento no aconselhamento e testes genéticos; a sua decisão não é baseada unicamente nas suas necessidades e

preferências, mas resulta de um processo psicossocial, recursivo, que envolve sentimentos de responsabilidade e acordos explícitos ou tácitos entre elementos da família, e também a consideração de outros significativos.

O quadro emocional da responsabilidade genética é clarificado através de estudos que descrevem autorresponsabilização e culpa no contexto do risco genético (Arribas-Ayllon, Sarangi, & Clarke, 2008), enquanto outros indicam a "culpa do sobrevivente", expressa por familiares que *escaparam* à herança de mutações genéticas deletérias (Cameron & Muller, 2009). Sentimentos de pressão familiar para a realização de testes genéticos, de solidariedade para com outros familiares que tenham realizado o teste, ou a possibilidade de poder evitar a transmissão de genes mutados às gerações futuras são descritos como os mais evidentes (Paneque, 2008). Sobel e Cowan (2000) referem a decisão de realizar testes genéticos, no contexto da doença de Huntington, como um "teste" à lealdade familiar, num processo em que os elementos da família julgam e avaliam a adesão a crenças, valores e regras da família. Ainda que pragmaticamente as provas genéticas sejam efetuadas através da recolha de uma amostra sanguínea individual para posterior análise laboratorial do ADN, a decisão de as realizar, e o seu resultado, têm profundas implicações nos outros membros da família e nesta enquanto sistema (Paneque, 2008; Patenaude, 2005; Vadaparampil, Wey, & Kinney, 2004; Van Riper & Gallo, 2005). Finkler (2000), num estudo de pesquisa antropológica com famílias norte-americanas com historial médico de cancro da mama e ovários, descreveu o sentimento de crescente controlo sobre o futuro associado à realização de testes genéticos, num processo em que o "destino familiar" é "contornado" através do envolvimento em práticas médicas de vigilância e profilaxia.

As repercussões psicológicas e emocionais do risco genético encontram-se proficuamente documentadas (Evers-Kiebooms *et al*., 2000; McAllister *et al*., 2007; Meiser, 2005; Patenaude, 2005; Power, Robinson, Bridge, Bernier, & Gilchrist, 2011; Schlich-Bakker, Ten Kroode, & Ausems, 2006). Do ponto de vista emocional, os estudos sugerem maior vulnerabilidade ansiógena e depressiva: a ansiedade surge associada a cenários de incerteza que rodeiam o risco acrescido a desenvolver cancro e ao facto de ser uma doença potencialmente fatal, prevalecendo sentimentos de preocupação, devastação e medo; a depressão advém de vivências de perda, incerteza e sofrimento psíquico prolongado, que tendem a desencadear menor participação e investimento social. O sofrimento psicológico em contextos de risco oncogenético pode ser prevenido através da identificação de indivíduos mais vulneráveis e subsequente provisão de intervenções específicas de apoio psicossocial. O preditor mais significativamente associado a dificuldades de ajustamento psicológico é o nível de stress psicológico pré-teste; outros preditores são a idade jovem, baixo nível sócio-cultural, género e presença de filhos (Van Oostrom, 2006).

De uma forma geral, os estudos sobre o impacto psicológico associado aos testes de suscetibilidade oncogenética focam-se em medidas de stress psicológico, antes e após a colheita de sangue e em períodos temporalmente variáveis uma vez conhecidos os resultados. Verifica-se a tendência para os indivíduos apresentarem níveis de ansiedade elevados e níveis depressivos baixos antes da colheita sanguínea para análise; com o conhecimento dos resultados do teste molecular, os níveis de ansiedade descem nos indivíduos não-portadores e tendem a aumentar nos portadores, que se apresentam também com maiores índices depressivos comparativamente com os não-portadores. Verifica-se a tendência destes dois níveis decrescerem em ambos os grupos de indivíduos durante os meses seguintes; após um ano, ambos os grupos apresentam níveis idênticos de ansiedade e depressão, exceto quanto à preocupação face ao cancro (mais elevado em indivíduos portadores) (Van Oostrom, 2006).

Uma questão presente na literatura é a hipótese de maior vulnerabilidade psicológica de indivíduos pertencentes a famílias com historial de cancro, comparativamente à população geral. Alguns estudos indicam que entre 20 e 30% de indivíduos com historial familiar sugestivo (afetados e não-afetados pela doença) reportam níveis significativos de stress psicológico, que no caso de resultados inconclusivos adquirem uma expressão ainda mais significativa (Butow *et al*., 2005; Power, Robinson, Bridge, Bernier, & Gilchrist, 2011). Outros estudos indicam, contudo, níveis semelhantes ou até inferiores comparativamente aos obtidos na população geral (Arver, Hagermark, Platten, Lindblom, & Brandberg, 2004), apontando a experiência prévia com a doença na família como um factor protector do ajustamento psicológico. Esta evidência verificou-se também em estudos no contexto de doenças neurológicas de início tardio (Paneque, 2008). Por outro lado, Santos, Figueiredo, Gomes e Sequeiros (2010) reportaram um acréscimo de ansiedade perante a morte e a diminuição do sentimento de imortalidade simbólica em familiares (com e sem risco acrescido) de indivíduos diagnosticados com polineuropatia amiloidótica familiar I (PAF-I).

Os estudos que referem o funcionamento familiar subjacente ao teste de suscetibilidade e risco oncogenético escasseiam. Van Oostrom (2006) descreve, num estudo prospetivo com indivíduos que realizaram o teste de suscetibilidade oncogenética para cancro da mama e ovários e cancro colorectal, indicadores específicos do funcionamento familiar dos sujeitos com níveis mais elevados de stress psicológico e preocupação com o cancro. Os resultados apontaram para: comunicação intrafamiliar sobre o risco de cancro hereditário inibida, funcionamento da família nuclear percebido como caótico e rígido, escasso suporte emocional do cônjuge e baixos níveis de diferenciação em relação à mãe. Keenan e colaboradores (2004) descreveram o contexto familiar de mulheres que realizaram teste molecular para *BRCA*1/2, indicando níveis elevados de coesão e expressividade emocional e predominantemente orientados para o crescimento pessoal e independência.

Um estudo recente que envolve uma amostra considerável em países escandinavos sugere que indivíduos envolvidos no aconselhamento oncogenético não apresentam níveis de ansiedade e stress mais elevados, quando comparados aos de indivíduos envolvidos noutros procedimentos em oncologia médica (como a realização de segundas mamografias devido a resultados suspeitos em exames anteriores ou receber um diagnóstico de cancro) ou da população geral (Roshanai *et al*., 2011). Os resultados apontam o efeito benéfico do aconselhamento oncogenético no ajustamento psicológico dos consultandos.

Alguns estudos qualitativos especificam uma ligação da vulnerabilidade psicoafetiva a fatores relacionais, como: ambivalência face ao desempenho de papéis familiares (Kenen, Ardern-Jones, & Eeles, 2006), preocupação pelo eventual impacto negativo do estado emocional nos outros significativos (Di Prospero *et al*., 2001) e ansiedade em relação a falar com os familiares sobre o risco genético (Hamilton, Bowers, & Williams, 2005). Relativamente aos aspetos psicológicos, a designação de Lopes e Fleming (1998), "a trama subterrânea intergeracional", usada num estudo no contexto da PAF-I, expressa com acerto alguns aspetos psicológicos subjacentes às doenças genéticas – assunção também sugerida por Brouwer-Dudokde e colaboradores (2002) a propósito da doença de Huntington: sentimentos de culpa associados à transmissão de genes mutados aos descendentes; e complexos sentimentos de perda, quer na realidade externa (familiares, integridade física associada à vivência corporal mutilada ou fragmentada), quer no plano interno (identidade enquanto indivíduo saudável; autoestima, autonomia).

As implicações psicoafetivas descritas enquadram-se no espectro do risco crónico (Kenen, Ardern-Jones, & Eeles, 2003) e da disrupção biográfica, articuladas com um sentimento de desamparo existencial e de rutura da continuidade entre passado, presente e futuro e manifestando-se na esfera identitária e das relações sociais (Bury, 1982; Karnilowicz, 2011). Vários estudos descrevem sentimentos de isolamento em mulheres portadoras de mutações *BRCA*, sobretudo em mulheres a quem foram realizadas mastectomias profiláticas (Kenenet *et al*., 2006), que se traduzem em barreiras percebidas entre as próprias e os outros ("separação social"), ativando sentimentos de solidão e de distanciamento face a outros significativos (Foster, Eeles, Ardern-Jones, Moynihan, & Watson, 2004).

### 4.3. O gene no sistema familiar: cenários de atuação

O funcionamento familiar face às condições crónicas de doença tem sido conceptualizado a partir da noção de crise, um acontecimento que perturba o equilíbrio do sistema, ameaçando a sua sobrevivência e exigindo reajustes no seu funcionamento (Alarcão, 2000). O caráter crónico das exigências da doença incorpora interseções com o ciclo de vida individual e familiar e suas tarefas desenvolvimentais normativas (Boss, 2001). As doenças hereditárias afetam o sistema familiar de

modo transgeracional e expõem as famílias a crises mais ou menos agudas, temporalmente prolongadas ou circunscritas, com graus variáveis de incerteza e de efeito cumulativo. A vivência subjetiva das doenças (e do risco) no sistema familiar constrói-se na interdependência espacio-temporal das relações biológicas e intergeracionais, expandindo assim a "ótica molecular" associada à identidade genética. As distintas determinações sociais, culturais, morais ou processuais dos sentimentos face à doença hereditária e ao significado da informação genética podem assumir contornos contraditórios ou ambíguos (Góngora, 2004) ou funcionar como uma "lente de aumento" que redimensiona os processos familiares, em particular os emocionais (McDaniel, Kepworth, & Doherty, 1992).

A resposta de indivíduos e suas famílias aos desafios das doenças genéticas pode ser compreendido através das dimensões normativas do funcionamento familiar (McDaniel, Rolland, Feetham, & Miller, 2006a; Peterson, 2005; Walsh, 1993): i) organização e estrutura das relações familiares; ii) padrões e processos comunicacionais; e iii) sistema de crenças da família; iv) padrões intergeracionais de confronto com as dificuldades e crises (*coping*); e v) desafios específicos da fase do desenvolvimento familiar (McGoldrick & Carter, 1999). As famílias, perante a (ameaça da) doença hereditária, efetuam movimentos de acomodação entre o stressor e a sua cultura de gestão da adversidade. Este processo insere-se num *continuum* temporal e desenrola-se através de um sistema de regras, tabus, expectativas, significados, valores e, em particular, modos de gerir o sucesso e a perda, padrões de lidar com o risco e a resiliência, desafios e mudança (Boss, 2001).

A justaposição de várias especificidades inerentes às doenças hereditárias e o ciclo vital individual e familiar pode determinar alterações nas suas transições desenvolvimentais normativas. Uma perspetiva desenvolvimental do ciclo de vida envolve uma progressão normativa sequencial ao longo de vários estádios, com tarefas e mudanças psicossociais antecipadas (McGoldrick & Carter, 1999; Relvas, 1996). As mudanças associadas à linha temporal das doenças genéticas (Street & Soldan, 2000; Rolland & Williams, 2005), à perceção do risco decorrente da confirmação do estatuto genético ou a mudanças físicas devido à emergência de sintomas, potenciam a perda antecipatória de expectativas individuais e familiares em relação a projetos e planeamento futuros (Rolland, 2006).

Rolland (1994) adota o modelo de Combrick-Graham (1985, *in* Rolland, 1994) na compreensão do desenvolvimento da doença, dos indivíduos e das famílias. O modelo descreve o ciclo de vida da família em espiral, contemplando um sistema familiar de três gerações que oscila no tempo entre períodos de elevada coesão familiar (centrípetos) e períodos de baixa coesão (centrífugos). Os períodos centrípetos e centrífugos implicam um encaixe entre tarefas das fases de desenvolvimento familiar e a necessidade relativa dos membros da família em ajustarem o seu funcionamento individual entre o interior e o exterior da família. Nos períodos centrípetos, a família concentra-se

no seu interior, havendo uma demarcação mais clara dos limites exteriores, enquanto se diluem os que medeiam os indivíduos. Nos momentos centrífugos é enfatizada a relação com o exterior, a estrutura muda para se acomodar às tarefas da vida fora da família. Estes conceitos permitem relacionar os ciclos vitais da doença, com os dos indivíduos e os das famílias. Na generalidade, as doenças graves, como os cancros, implicam movimentos centrípetos no sistema familiar, semelhantes à chegada de um novo membro. Os procedimentos inerentes à monitorização do risco, as necessidades e adaptações ou a aquisição de novos papéis relacionados com a gestão da saúde e da informação genética, os sinais e sintomas e o medo da perda requerem que a família se debruce para o seu interior. Esta orientação provoca ansiedades normativas distintas dependendo do nível de desenvolvimento da família e dos indivíduos.

A informação genética e contexto de risco que lhe está subjacente, ou de doença, desafia vários aspetos da organização familiar exigindo a revisão de objetivos e prioridades instrumentais, emocionais e relacionais, a adaptabilidade e permutação de papéis, hierarquias e poder, gestão da proximidade e distância e reconfiguração da identidade familiar "saudável" (Góngora, 2004). Penn (1983) descreveu as repercussões das condições crónicas de doença no sistema familiar, destacando as coligações que envolvem o elemento doente e o(a) cuidador(a) primário e fenómenos de exclusão emocional. Evans (2006), a partir das implicações das teorias da vinculação na psicologia da família e terapia familiar, nota que nas famílias em que as doenças hereditárias são mais incidentes, existe a expectativa de repetição de perdas similar às experienciadas ao longo das gerações anteriores. Para além da potencial rigidificação identitária da família face às exigências da doença (Patterson & Garwick 1994), a experiência da perda antecipatória (Rolland, 2006), com efeito, inibe o desenvolvimento de bases de vinculação seguras, em particular nas famílias em que estas circunstâncias se repetem ao longo de gerações.

Adicionalmente, o caráter intergeracional da perda ameaça a adequada diferenciação emocional face às figuras parentais. Esta dificuldade é suscetível de perturbar a gestão da separação-autonomização, sobretudo em etapas do ciclo vital cujas tarefas normativas envolvem maior abertura familiar à socialização e individuação. A dinâmica interdependente entre as exigências da doença e a rede de cuidados informais (e o impacto emocional que se lhe associa) pode potenciar, no contexto de uma rigidificação estrutural e organizacional do sistema familiar, o progressivo isolamento social. Da gestão equilibrada da crise podem emergir significados da doença funcionais e coerentes com formas de funcionamento familiar resilientes que possibilitem a continuidade do desenvolvimento da vida familiar, evitando, assim, o seu bloqueio evolutivo.

O sistema de crenças da família desenvolve-se a partir de experiências e tradições partilhadas entre os membros, que sustentam modelos explicativos sobre determinado evento ou condição. As famílias tendem a alocar papéis aos seus membros, como "guiões" (*scripts*) familiares, traduzindo

as construções, histórias e crenças sobre as relações familiares ao longo de gerações e que refletem a sua interpretação sobre a sua história no confronto com a realidade (Reiss, 1981). As crenças dão sentido e orientação à vida familiar facilitando a continuidade entre passado, presente e futuro; o conjunto de crenças sobre a saúde e a doença influenciam as atitudes e os comportamentos na gestão da doença. No caso das doenças genéticas, estigma, controlo e competência percebidos sobre as causas e exigências da doença são aspetos relevantes na adaptação individual e familiar à informação genética e ao processo de aconselhamento genético. Tipicamente, as crenças de uma família em relação à doença e ao eventual envolvimento em medidas profiláticas são permeadas por crenças religiosas (Rolland & Williams, 2005).

No contexto da doença de Huntington, Kessler e Bloch (1989) descreveram o processo de pré-seleção, através do qual um elemento da família (tipicamente da geração mais nova) é selecionado como quem mais provavelmente irá desenvolver a doença. Trata-se de um processo fundado em semelhanças físicas ou atitudinais com o indivíduo afetado aquando do aparecimento dos primeiros sintomas, de caráter inconsciente, e que atenua a ansiedade inerente à incerteza quanto à eclosão da doença. Werner-Lin (2007) descreve um processo semelhante em famílias com elevada suscetibilidade a cancro da mama e ovários. O conceito de projeção familiar foi proposto por Bowen (1978, *in* Reiss, 1981) para designar os padrões de transmissão transgeracional na família, através dos quais conflitos não-resolvidos, papéis e tarefas são perpetuados através das gerações. Os padrões multigeracionais da doença influenciam a forma como os indivíduos perspetivam o seu legado biológico (Hunter & Rowles, 2005; Walsh & McGoldrick, 1988). Um diagnóstico oncológico pode ser um evento atormentador na família devido ao seu impacto individual, relacional e simbólico (Kleinman, 1988; Sontag, 2009). Um outro exemplo é o padrão de respostas e processos desencadeados na dinâmica familiar quando um indivíduo atinge a idade com que elementos significativos de gerações anteriores foram diagnosticados com a doença ou experienciaram momentos marcantes na sua trajetória de confronto com a mesma.

### 4.4. Redes familiares e sociais e saúde: ajuda comunal, construção narrativa e influência social

O estudo das redes familiares e sociais no âmbito da adaptação às doenças hereditárias tem sido apontado como uma linha de investigação importante na provisão de serviços psicossociais no aconselhamento genético. Alarcão e Sousa (2007: 357) sintetizam as várias definições de rede social pessoal, elucidando que estas traduzem *"sistemas abertos que, através de um intercâmbio dinâmico entre os seus membros e os elementos de outros grupos sociais, potencializam outros recursos (cada membro de uma família, grupo ou instituição enriquece-se através das múltiplas relações que cada um dos membros desenvolve)"*.

Como o risco genético de contrair uma doença pode abranger vários familiares, o conceito de ajuda (*coping*) comunal tem sido proposto como quadro de referência teórico nalguns estudos e sugerido como particularmente relevante para fazer face às exigências psicossociais das doenças hereditárias (Koehly *et al*., 2008). Este conceito pressupõe que os membros da família ou do grupo alargado, perante a perceção de um stressor comum, comuniquem e desenvolvam ações coletivas para ultrapassarem a situação. Em famílias com elevada suscetibilidade genética a cancros hereditários, esforços cooperativos na rede relacional intrafamiliar podem revelar-se um recurso decisivo na gestão bem sucedida da saúde. Por exemplo, destacam-se, na perspetiva de suporte instrumental, a disseminação da informação acerca do risco genético pelos familiares (que poderão beneficiar de aconselhamento genético), a provisão de apoio informativo e acesso a contactos de profissionais de saúde ou de centros clínicos. O encorajamento ao envolvimento em medidas de vigilância e monitorização do risco, incluindo a realização de testes genéticos, emerge como importante função de suporte emocional. A ajuda comunal designa, assim, o processo coletivo, recíproco e interativo, em que famílias e grupos se envolvem para enfrentar uma ameaça percebida como sendo comum aos seus membros (Lyons, Mickelson, Sullivan, & Coyne, 1998).

Estudos qualitativos que focam a adaptação psicossocial ao risco genético têm enfatizado o papel da construção narrativa (Sanders, Campbell, Donovan, & Sharp, 2007). As narrativas contribuem para a criação de significado e de identidade; organizam a vida familiar através da manutenção de estabilidade e conexão entre os elementos; enfatizam significados partilhados e potenciam a expressividade emocional ao longo das gerações (Hoffman, 1990). Narrativas intergeracionais ativam a continuidade simbólica pela invocação de mitos familiares que moldam o comportamento e a interação familiar. O acesso ao passado da vida familiar através da história médica familiar no contexto do aconselhamento genético é fundamental. A construção narrativa por via do conhecimento experiencial adquirido no contexto das vivências intergercaionais tem sido um aspeto que a investigação sugere como relevante na perceção e gestão familiar do risco, tomando precedência face a estimativas objetivas provenientes do encontro médico (D'Agincourt-Canning, 2005). A vivência familiar da doença enforma histórias de sofrimento e incerteza que perpassam as gerações. Perante contextos familiares fortemente pautados por incerteza e ambiguidade (*e.g.*, aguardar por resultados de testes genéticos ou a tomada de decisões para a realização de cirurgias profiláticas), a criação de uma narrativa coerente e funcional entre passado, presente e futuro pode ser relevante para o fortalecimento da identidade familiar e uma gestão harmoniosa da incerteza face ao futuro, nomeadamente na criação de significado para a vulnerabilidade.

A pespectiva sistémica da família enfatiza que indivíduos, seus familiares e outros membros da sua rede social pessoal integram um contexto social onde as trocas relacionais entre estes intervenientes se processam reciprocamente. A estreita ligação entre vínculos interpessoais e a saúde física e

mental tem sido vastamente estudada, destacando-se o papel da influência social e comunicação interpessoal nos comportamentos para a saúde (Berkman, Glass, Brissette, & Seeman, 2000). A extensão das trocas de recursos a nível intrafamiliar e social influencia o bem-estar emocional e o modo como os indivíduos agem sobre a gestão da saúde, inclusive no contexto de doenças genéticas (Kenen *et al*., 2006; Werner-Lin, 2008). Vários estudos com famílias afetadas por cancros hereditários têm demonstrado que fatores como o apoio emocional, acesso a informação e encorajamento para a deteção precoce e medidas preventivas assumem uma importância decisiva na adaptação psicossocial de indivíduos e famílias (Ersig, Williams, Hadley, & Koehely, 2009; Hughes *et al*., 2002; Koehly *et al*., 2003; 2009; McCaan *et al*., 2009) e influenciam a decisão de envolvimento no aconselhamento genético (Peterson, Watts, & Koehly, 2003).

A importância dos aspetos relacionais extravasa as redes familiares; o papel de outros significativos em mulheres saudáveis envolvidas no aconselhamento genético para cancro da mama e ovários foi destacado por Kenen, Ardern-Jones e Eeles (2004) como um recurso de apoio social relevante. As autoras avançam a ideia de que este "espaço inter-relacional" alternativo à família adquire maior importância em famílias cuja capacidade de expressão emocional é limitada.

### 4.5. Intervenções com famílias

A literatura tem demonstrado a influência das relações familiares na saúde, embora não seja claro quais os mecanismos implicados neste processo. Weihs, Fisher e Baird (2002) reviram extensamente a literatura sobre famílias e saúde, identificando fatores protetores e de risco do funcionamento familiar para a saúde dos seus elementos. Proximidade e mutualidade das relações familiares, competências do cuidador e comunicação aberta sobre a doença, foram os fatores protetores descritos; conflitos, isolamento, stressores externos, disrupção das tarefas desenvolvimentais, dificuldades psicológicos decorrentes da doença e rigidez estrutural, foram apontados como os fatores de risco.

As intervenções dirigidas a famílias no contexto de doenças físicas ou incapacidades crónicas acompanham a premissa da sua influência mútua e bidirecional. Ou seja, de que as doenças têm impacto no sistema familiar como um todo e que este pode influenciar negativa ou positivamente a saúde do membro afetado (McDaniel, Hepworth, & Doherty, 1992). As intervenções familiares podem melhorar o funcionamento familiar e o bem-estar e saúde emocional dos seus membros. Os benefícios centram-se sobretudo na prestação de apoio emocional e na facilitação de um sentimento de pertença; relações familiares percecionadas como negativas e hostis têm um efeito mais poderoso na saúde do que as relações familiares suportivas (Campbell, 2003).

McDaniel, Rolland, Feetham e Miller (2006b) preconizam um modelo geral de prevenção piramidal fundado em premissas universais de apoio psicossocial centrado na família, movendo-se

depois, de acordo com os fatores de risco identificados e perante dificuldades persistentes, até intervenções de monitorização e de enfoque clínico-terapêutico. O modelo pressupõe o envolvimento multidisciplinar de profissionais de saúde com formação específica na área do risco genético, incluindo terapeutas familiares e técnicos de saúde mental. Os autores agrupam as intervenções psicossocias com famílias no contexto da gestão da informação e do risco genético em duas grandes categorias, cuja delimitação comporta sobreposições: intervenções psicoeducativas (em formato unifamiliar ou em grupos de discussão multifamílias) e intervenções de caráter terapêutico (terapia familiar, terapia de casal e psicoterapia individual). Tais intervenções requerem flexibilidade quanto às orientações teóricas e técnicas empregues (cognitivo-comportamentais, psicodinâmicas e sistémicas), embora mantendo o enfoque familiar e o seu caráter breve[23].

Os objetivos gerais das intervenções familiares face a condições crónicas de doença, cuja conceptualização teórica deriva do paradigma normativo sistémico da saúde e doença (Rolland, 1994), focam-se no reforço das competências da família na gestão das exigências decorrentes da doença, neste caso, da suscetibilidade genética. De um modo geral, as intervenções visam capacitar indivíduos e famílias em áreas específicas da gestão da doença (ou do seu risco acrescido), como a compreensão da informação genética, gestão emocional face à incerteza e aos resultados dos testes genéticos, apoio à tomada de decisão, comunicação intrafamiliar, expansão da rede social pessoal e interação com o sistema de saúde. A acomodação das crenças familiares sobre a saúde (e em particular, da hereditariedade e do risco genético), a criação de um significado para a informação genética que permita a manutenção da identidade familiar e da continuidade funcional entre os eixos sincrónico e diacrónico do desenvolvimento familiar, e a maximização do apoio social são orientações clássicas no trabalho com famílias de um ponto de vista familiar-sistémico, ao que se acresce uma perspetiva colaborativa no contexto da equipa de cuidados técnicos e assistenciais (McDaniel, Hepworth, & Doherty, 1992).

## 5. ENFOQUE DA INVESTIGAÇÃO

Os desafios associados aos avanços da genómica no diagnóstico, tratamento e prevenção de doenças colocam a suscetibilidade acrescida a doenças, como os cancros hereditários, numa interface de contornos complexos entre o indivíduo, a família e os cuidados de saúde. A literatura sugere a tendência crescente de os cuidados de saúde na era genómica assumirem um enfoque

[23] Embora, como os autores notam, no contexto de um *setting* psicoterapêutico em que o pedido inicial se centra nas dificuldades colocadas por uma doença genética ou pelo seu risco acrescido, possam emergir outras questões ou problemáticas cujo aprofundamento passe a merecer enfoque primordial numa intervenção mais prolongada.

familiar, traduzindo na prática a centralidade que a família ocupa no aconselhamento genético e a necessidade de aprofundar o conhecimento sobre os diferentes aspetos do contexto intrafamiliar inerente a gestão do risco e aconselhamento genético (Peters, Djurdinovic, & Baker, 1999).

Perseguindo as premissas do paradigma sistémico da saúde-doença (Rolland, 1994), esta investigação tem como finalidade contribuir para o conhecimento da experiência individual e familiar do risco e aconselhamento oncogenético e de como esta pode ser contemplada no desenvolvimento de intervenções de apoio e na prestação de cuidados de saúde genómicos. A escassez de intervenções de apoio psicossocial na área, dirigidas a famílias cujos membros estão ou podem estar em risco acrescido de desenvolver cancro, motivou o desenvolvimento de objetivos específicos, que a seguir se enumeram: i) caracterizar a experiência psicossocial individual e familiar do aconselhamento oncogenético; ii) desenvolver, implementar e avaliar uma intervenção de cariz psicoeducativo multifamiliar no risco oncogenético; e iii) auscultar a perspetiva dos profissionais de saúde quanto à incorporação de apoio psicossocial no aconselhamento oncogenético. Na prossecução destes objetivos foram usados pressupostos da investigação-ação e metodologias qualitativas de recolha (*focus group*, entrevistas) e de análise de dados (*grounded theory*). A investigação incluiu o desenvolvimento de estudos interligados, que a seguir se apresentam, acoplados em três capítulos, a partir dos quais de organiza o desenvolvimento desta tese.

Esta investigação procurou, assim, não somente desvelar caminhos científicos relevantes mas também potenciar formas de atuação e colaboração entre vários domínios com impacto na qualidade de vida das populações que foram objecto de estudo.

### 5.1. Aconselhamento oncogenético: experiência individual e familiar

O capítulo I engloba dois estudos que procuraram descrever a experiência individual e familiar do aconselhamento oncogenético. Ambos os níveis de experiência estudados assumem relevância no aprofundamento de significados e determinantes individuais, relacionais e familiares associados ao risco e aconselhamento genético. Os resultados procuram caracterizar as trajetórias de indivíduos e famílias em diferentes momentos da sua adaptação ao risco de doença e ao aconselhamento oncogenético, descrevendo as suas implicações na interface psicossocial entre os domínios médico, individual e familiar. Assim, descrevem-se especificamente as implicações instrumentais, emocionais e relacionais na gestão da saúde e no desenvolvimento individual e familiar. A caracterização efetuada enquadra as necessidades individuais e familiares associadas ao aconselhamento oncogenético e sugere perspetivas de pesquisa e recomendações práticas para a provisão de apoio psicossocial.

### 5.2. Intervenção psicoeducativa: abordagem multifamiliar

O segundo capítulo descreve o desenvolvimento, implementação e avaliação de uma intervenção psicoeducativa em dois grupos de discussão multifamílias. A psicoeducação é considerada uma intervenção-chave em oncologia psicossocial, integrando a educação para a saúde e apoio psicossocial numa perspetiva multidisciplinar. No âmbito das intervenções familiares sistémicas, os grupos de discussão multifamiliares conheceram um crescente reconhecimento como forma de potenciar a resiliência familiar face a condições crónicas de doença (Gonzalez, Steinglass, & Reiss, 1989; McDaniel *et al*., 2005), incluindo o risco genético (McDaniel *et al*., 2006b).

Foram implementadas duas intervenções multifamiliares com grupos homogéneos quanto ao tipo de suscetibilidade oncogenética, envolvendo indivíduos e seus familiares em risco de cancro da mama e ovários e em risco de cancro colorectal. Foi realizado um *focus-group* para avaliar o impacto psicossocial e prático e recolher sugestões de melhoria da intervenção tendo em conta a experiência de participação. Os resultados incidem sobre o impacto do programa segundo os participantes, sugerindo também temas específicos para melhorar os guiões de intervenção.

### 5.3. Incorporação de apoio psicossocial no aconselhamento oncogenético: perspetivas dos profissionais

O terceiro capítulo acopla dois estudos sobre a perspetiva de profissionais do aconselhamento genético quanto à sua prática e ao modo como o apoio psicossocial a indivíduos e famílias é disponibilizado no contexto do sistema de saúde. Especificamente, num dos estudos, os profissionais de saúde foram auscultados em relação às potencialidades e limitações da intervenção multifamiliar descrita no capítulo II, bem como quanto à pertinência e viabilidade da sua incorporação nos serviços de genética (*Are family-oriented interventions in Portuguese genetics services a remote possibility? Professionals' views on a multifamily intervention for cancer susceptibility families*). Este estudo procurou fiabilizar os dados obtidos na avaliação feita pelos participantes à intervenção através da triangulação das fontes dos dados qualitativos (Patton, 1990). Noutro estudo, os profissionais de saúde refletem acerca dos desafios associados á prática do aconselhamento oncogenético e descrevem as necessidades atuais dos serviços de saúde para a incorporação de intervenções de apoio psicossocial a indivíduos e famílias no contexto do aconselhamento oncogenético (*Challenges for cancer genetic counselling: a qualitative study in Portuguese oncogenetic services*). Os resultados obtidos proporcionam uma base exploratória de implicações para o planeamento e prática do aconselhamento oncogenético.

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# CAPÍTULO I

## ACONSELHAMENTO ONCOGENÉTICO: EXPERIÊNCIA INDIVIDUAL E FAMILIAR

# 1. EXPERIENCING GENETIC COUNSELLING FOR HEREDITARY CANCERS: THE CLIENT'S PERSPECTIVE[24]

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## ABSTRACT

As genetic health care expands and genetic testing becomes more widely available, it becomes relevant to understand how individuals involved in genetic counselling are integrating this new information in health management and into their lives. This article examines the client's experiences of genetic counselling for hereditary cancers, which definitely play a major role in the assessment of their needs and also lead to improvement of the psychosocial focus in genetic counselling protocols. Methods include a semi-structured interview, administered in two focus groups, comprising 10 (5 + 5) participants after attending genetic counselling for hereditary cancers at a Portuguese public hospital. Findings suggest an experience embedded in two dimensions: i) instrumental (goals, needs and decision making); ii) emotional (uncertainty regarding genetic risk and an emotional complex). Uncertainty plays a crucial role, especially in two moments: i) the hiatus between genetic testing and knowing its result; and ii) after being confirmed as carrying a cancer susceptibility gene mutation. The spectrum of genetic illness comprises an intensely complex emotional experience that challenges individuals and their families in terms of health management, and personal and family planning. Recommendations are included in order to enhance the services available by expanding psychosocial support.

**Keywords:** genetic counselling, hereditary cancers, psychosocial genetics, focus-group.

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## 1.1. INTRODUCTION

Genetic testing is currently available for a vast number of conditions, namely for diverse hereditary cancers, allowing for prediction and prevention to be made before illness progression. Breast and colorectal cancer are the most common forms of hereditary cancers (Schneider, 2002). It is estimated that 5-10% of cancer cases are due to inherited mutations. However, if a known cancer germline mutation is present, the risk may increase to 80%, although variable penetrance remains uncertain (Hodgson et al, 2007). Cancer risk counselling has grown rapidly in recent years to become a major area of specialisation within genetic counselling. Its main purpose is to evaluate the tumour risk in families whose members have a hereditary predisposition for cancer and to inform them about treatment and preventive strategies, ensuring they have the ability to make informed decisions (National Society for Genetic Counselors, 2006; Sifri et al, 2004).

The typical guidelines for genetic counselling for hereditary tumours (Trepanier et al, 2004) are as follows: a) collection of family and individual medical history; b) psychosocial and genetic risk assessment; c) predictive testing for genetic susceptibility to cancer; and d) surveillance and preventive screening, treatment and follow-up sessions. These procedures were followed at the genetic centre where our study took place (Hereditary Tumours Consultation, Centre for Medical Genetics and Human Reproduction at the University Hospital of Coimbra, Portugal).

Clients and their families also manifested an intense biopsychosocial experience while going through this process. Rolland and Williams (2006) expanded the Family System Illness Model (Rolland, 1994) to incorporate genetically influenced disorders and provide a framework for that experience. The original post-clinical onset model conceptualised chronic illness as an ongoing process framed in three phases (Rolland, 1994): the crisis phase, which includes the initial contact with illness symptoms, treatments and institutional settings, and family flexibility; the chronic phase, which illustrates how the illness affects daily activities, where the task of keeping the balance between personal and family needs and illness constraints assumes vital importance; and the terminal phase, which is dominated by the impending loss and the need for family reorganisation. As the potentialities of genetic counselling posed a challenge for the original model, the authors decided to expand their initial approach so that genetic disorders could also be included in that model. Rolland and Williams (2006) have thus included the time before the illness clinical onset: the nonsymptomatic time phases of genomic disorders. These include four different phases: i) awareness of possible genetic risk – this phase involves seeking basic information about the illness and establishing initial communication in the family; ii) crisis I (pre-testing phase) – this phase encompasses the psychosocial understanding of the illness, the active consideration and decision regarding testing, and the information of family members; iii) crisis II (test/post-testing phase) – this phase involves the incorporation of the testing outcome into personal and family life,

and the consideration of the prophylactic treatment options available; iv) long-term adaptation (if results are positive) – this phase refers to the balance between proactive personal and family planning and the need for up-to-date genetic information. The updated model includes the experience of genetic disorders and genetic counselling, but this process still needs to be further examined in order to achieve deeper understanding of specific genetic disorders (as hereditary cancers). This is crucial to the development of adequate psychosocial support for those going trough genetic counselling. Although research shows that genetic testing for hereditary cancer risk is a distressing experience which causes significant emotional burden for clients and their families (Meiser, 2005; Patenaude, 2005; Shiloh et al, 2009), few services are able to provide psychosocial support during the genetic counselling process (Speice et al, 2002; Werner-Lin, 2008).

Therefore, this study examines how individuals experience genetic counselling for hereditary cancers. Findings will provide a series of guidelines intended to enhance genetic counselling procedures, increasing understanding of the psychosocial needs of clients and their families.

## 1.2. METHODS

This study uses the qualitative method known as the focus group, which is a form of group interviewing. This was the method chosen because (Krueger and Casey, 2000; McLachlan, 2005): it is pertinent for exploratory approaches, since it is a rich method of revealing experiences and perceptions; it is appropriate for sensitive topics, which is the case of genetic counselling experience, as it provides a safe environment for participants to share their thoughts and feelings. The focus group was used in place of one-on-one in-depth interviews because the group setting allows individuals to use the ideas of others as cues to more fully elicit their own views (Cohen, Manion and Morrison, 2000). Although focus group results are limited in terms of significance, they can be useful for identifying issues which need to be further examined in a large representative sample of the population using other research techniques (McLachlan, 2005). Ethical approval for this study has been granted by the Ethics Committee of the University Hospital of Coimbra (Coimbra, Portugal).

### 1.2.1. Procedures

It was decided to organise constructed and homogeneous focus groups (McLachlan, 2005; Patton, 1990). Constructed groups (where the participants have not met before) can be useful to minimise potential for group conformity. Participants who are unlikely to meet again face less personal cost if they express divergent views and are likely to be more honest. Homogenous groups (those whose members share an experience) are more likely to yield rich data than groups with little in common. Therefore, the following inclusion criteria were used: participants should have attended at least one

genetic counselling consultation; living at a reasonable driving distance from the hospital (less than one hour); the group should include participants who were at different moments on the genetic counselling timeline, and who showed both positive/negative genetic test results; participants should belong to different family groups.

Participants were recruited according to the following process: potential participants were given a brief description of the research project at the genetic counselling consultation by the doctor, who had been previously informed about the inclusion criteria, and were thus asked to participate in the study. Those who agreed to participate (10 participants) were then introduced to the researcher (first author), who explained the objectives of the study, as well as the group setting and the data management procedure, assuring confidentiality and anonymity. An interview was scheduled to collect demographical and psychosocial data, and an informed consent form was signed. Finally, telephone contact was made to deal with practical issues, such as the focus group schedule. We decided to run 2 focus groups (5 + 5 participants) for practical reasons (according to participants' personal agendas), and especially to strengthen the validity and reliability of data (McLachlan, 2005).

The interviews were applied according to the guidelines for conducting focus groups (Piercy and Hertlein, 2005), with each interview lasting approximately two hours. An experienced researcher (first author) moderated the interview, which was audiotaped with the permission of the participants. After a brief introduction to the focus group process, a short list of core questions was used as an interview guide. Questions were formulated using the semi-structured interview method, and were based on the existing literature on cancer genetic counselling and its psychosocial impact (Rolland and Williams, 2006) (Tab. 1.1.). By using open-ended questions, the moderator created and promoted a conversational atmosphere which encouraged discussion among participants. Some prompts were delivered if the participants had difficulty talking about their experience and its meaning. At the end of the interview the moderator presented an oral summary and encouraged participants to give feedback on the focus group.

**Table 1.1. Focus Group Script**

| Topics | Questions |
|---|---|
| Experiences | *What is the main utility of genetic counselling? What are its advantages and disadvantages?* |
| | *What is the meaning of genetic risk / being at genetic risk?* |
| | *Why did you decide to come to a genetic counselling session?* |
| Psychosocial needs | *What was the easiest / hardest moment?* |
| | *What is the ideal way for you to feel supported?* |
| | *What would increase your sense of wellbeing?* |

### 1.2.2. Participants

Participants ($n$ = 10) were selected from the Hereditary Tumours Consultation of the Centre for Medical Genetics and Human Reproduction of the University Hospital of Coimbra (Coimbra, Portugal), using a convenience sampling. There were 136 new cases of patients who came to the consultation in 2008, mainly urban Portuguese users (77% were females). 19-44 years old was the most frequent age group (62%), followed by 45-59 years old (22%).

All participants were over 18 years of age, white, and Portuguese. Concerning the time phases of the illness, most of the participants were non-symptomatic at the time of the focus group, with an identical distribution across the genetic counselling timeline (Rolland and Williams, 2006) (Tab. 1.2.).

**Table 1.2. Participants' timeline of genetic illness phases and genetic counselling protocol**

| **Participants' timeline of genetic illness phases and genetic counselling protocol** | ***n* = 10** |
|---|---|
| **Time phase of genetic illness** | |
| *Nonsymptomatic* | |
| Presymptomatic | 5 |
| Post-clinical onset (chronic phase - symptoms remission) | 4 |
| *Symptomatic* | |
| Post-clinical onset (chronic phase) | 1 |
| **Genetic Counselling timeline** | |
| *Crisis I* | |
| Pre-test counselling | 2 |
| *Crisis II* | |
| Waiting for results | 3 |
| Initial adjustment period | 2 |
| *Long-term adaptation* | |
| Living with genetic information | 3 |
| **Genetic testing results** | |
| Positive | 5 |
| Negative | 2 |
| Awaiting results | 3 |

### 1.2.3. Data analysis

The interviews were transcribed and submitted to content analysis, using a grounded theory approach, based on genetic testing for hereditary syndromes and on the psychosocial processes of genetic illnesses (Rolland and Williams, 2006; Hurley et al, 2006). Two independent researchers (first and last authors) transcribed the interview, and analysed the transcription aiming to develop a set of categories that could elicit accounts of participants' genetic counselling experiences. Then, the researchers met to compare and discuss their categorisation proposals through a process of

successive refinement, until agreement was reached. The judges decided to perform the data analysis of both focus groups together since data were similar. Finally, a list of categories and subcategories was created and presented as qualitative findings from the focus group research, including descriptive summaries illustrated by quotes from the raw data (see results).

## 1.3. RESULTS

Results suggest that the client's experience of genetic counselling comprises two major dimensions, focusing on personal and family levels: i) instrumental, centred on pragmatic elements ("defining goals for genetic counselling", "emergence of needs" and "health care decisions and illness management"); and ii) emotional, which permeates all the experience ("uncertainty facing genetic risk" and the "emotional complex" when facing genetic risk assessment).

### 1.3.1. Instrumental categories

These involve a temporal dimension of the client's experience throughout the process of genetic counselling: since the moment when clients are referred for genetics consultation, usually by their family doctors, they develop expectations and beliefs which are represented as goals in the process; after the first genetic counselling session, informative, procedural and psychosocial needs emerge, concerning health management decisions and their impact on relatives.

#### *1.3.1.1. Defining goals for genetic counselling*

When genetic counselling emerges as a possibility, the client's main goal is "to know if there is a genetic predisposition", both at the individual and family levels:

> *"I want to know if what happened to me is going to happen to my family, whether the odds of getting cancer are high…"*. [Beatriz[25], female, 35 years old]

Participants reported to have limited prior knowledge of genetic issues. Genetic counselling is perceived as something "complex and scientific", quite distinct from other medical consultations. Consequently, users feel the need to obtain useful information and medical counselling to plan and implement preventive actions. In particular, they are interested in personal health management, which emerges as a preventive goal, and is both individually and family focused:

> *"I came here to know more about cancer prevention and to know if it is possible to help other relatives who have the mutation …"*. [António, male, 23 years old]

Once the genetic risk has been ascertained, users tend to perceive genetic testing as a family commitment, a matter of dedication and responsability:

[25]All participants' names have been changed for confidentiality purposes.

*"If the chances of being considered at risk are real, to take the* [genetic] *test is a matter of citizenship and respect towards other family members"*. [Manuela, female, 48 years old]

#### *1.3.1.2. Emergence of needs*

Participants mentioned that informative, procedural and psychosocial needs arise during the process of genetic counselling. Informative needs include the access to medical information, namely regarding test results and post-test scenarios, in order to facilitate the progressive integration of the anticipated consequences (as a pathogenic mutation carrier or non-carrier) into practical life and emotional experience. Clients need information, so they can feel more capable of making informed decisions:

*"It is very important to have access to medical information before we know the test results; we should be informed about what is going to be done, what for, the possible scenarios and its implications"*. [Beatriz, female, 35 years old]

Participants also suggested the creation of an informative website. The use of simple language by professionals, free of technical jargon, is viewed as fundamental, since it facilitates clients' understanding of genetic concepts. Procedural needs are focused on the genetic counselling protocol, particularly pointing to the diminution of the temporal hiatus between the uptake of genetic testing and the disclosure of results. At the service where the study was developed one can expect, on average, a 3-month hiatus, because the tests are assured by external services and then sent back to the hospital:

*"The waiting period between the testing and the communication of results was too long… this is prejudicial to our own well-being because we're always thinking about it..."*. [Manuela, female, 48 years old]

Participants also suggested that collaboration between the family doctor and the genetic centre should be improved to promote a better sense of identification among the clients, their families and the medical team:

*"Perhaps the collaboration between the genetic counsellor and the family doctor should be improved; family doctors have a better knowledge of the patients and their families, and can antecipate their reactions"*. [Joaquim, male, 48 years old]

The psychosocial needs experienced during genetic counselling were emphasised by all participants. However, they also pointed out that these needs emerge particularly while waiting for the test results and after the disclosure of results (especially in a positive scenario):

*"I felt there was a lack of emotional and psychological support between the first session and the communication of my test results"*. [Manuela, female, 48 years old]

Participants made some recommendations to improve the process of genetic counselling: making group sessions available for sharing the subjective facets of the experience; and creating a blog for sharing doubts.

#### *1.3.3.3. Healthcare decisions and illness management*

Following eligibility confirmation to proceed in genetic counselling, clients need to decide whether or not to take the genetic test. Participants reported that access to information is essential for making an informed decision. However, they also disclose its distressing potential:

> *"Being informed is an advantage; the more we know, the better we cope with the illness. But this kind of knowledge might also bring about some concerns and anxiety…".* [Orlando, male, 27 years old]

After genetic testing, clients know whether or not they carry the mutated gene that increases cancer susceptibility. Decision-making is required in terms of preventive measures, mainly for mid-role genetic illness management:

> *"We need to make decisions based on clues and medical recommendations, provided during the consultation… surgery, treatments…".* [Isabel, female, 36 years old]

In the meantime, genetic counselling may promote healthy behaviours:

> *"There were things that I could do for my benefit: I've decided to start going for a walk and I tried to quit smoking…".* [António, male, 23 years old]

Participants reported that decisions are considered within the family context. A common and main task for clients is (to decide) to inform those family members whose genetic risk was noticed by the physician, and to enlighten them about the importance of genetic counselling. Such effort is often difficult, due to geographical distance, family conflict and/or because people feel unable to properly give information to relatives:

> *"The doctor told me that some of my relatives might be at risk and that I should ask them to do some tests, but in some cases we haven't spoken for years!".* [Isabel, female, 48 years old]

This can be a time-extended process, especially when family members have not reached the age at which genetic testing is appropriate:

> *"My biggest concern is that my daughters cannot get the genetic test done yet (10 and 6 years); would it be the best option to trouble them with this issue?".* [José, male, 50 years old]

Planning for the future assumes particular significance in post-test consultation(s), because test results usually lead to a redefinition of personal, familiar and professional values. Reproductive issues are a matter of particular concern, mainly for those younger clients considering parenthood.

### 1.3.2. Emotional categories

Emotional categories are present throughout all the genetic counselling protocol, having a strong impact on participants' lives, mostly when the test results are positive. Uncertainty facing genetic risk assessment and an emotional complex are the main features mentioned.

#### *1.3.2.1. Uncertainty facing genetic risk*

Uncertainty emerges after the referral to a cancer genetics centre (and sometimes previously, prompted by the awareness of the family cancer history), continues after the family medical history has been explored and lasts, at least, until the disclosure of genetic testing results. Uncertainty is also associated with a lack of knowledge about genetics:

> *"It is like an unfinished book where we are able to find more about ourselves as a person and as a genetic entity; we can't control it… I don't know whether I'm a mutation carrier or not"*. [Orlando, male, 27 years old]

Uncertainty levels increase while waiting for test results; uncertainty levels were found to decrease only in the case of a negative result:

> *"Uncertainty is central and it is always present… the risk rates are so variable!"*. [Beatriz, female, 35 years old]

#### *1.3.2.2. Emotional complex*

According to participants, several emotions emerge throughout genetic counselling, including anxiety, fear, guilt, stress and helplessness attached to a feeling of "loss of control". This emotional impact is particularly evident during the hiatus between genetic testing and the disclosure of results, and takes the form of an effect typically associated with genetic clinical procedures: the lack of control ("*I want to know what's going on but I'm feeling stuck"*):

> *"I was very anxious after the test and before the communication of results… I was always thinking about the test results… and it took so long! … There were moments when I just felt like dying…"*. [Nicolau, male, 68 years old]

While waiting for the test results, fear assumes a double meaning: i) the fear of having inherited a pathogenic mutation from their ancestors; ii) the fear of passing the mutated gene on to their existing and/or future descendants.

Participants revealed that those whose test results were negative felt a sense of relief; and those whose results were positive had different concerns, apparently depending on the illness phase: non-sympthomatic participants recognised strong anxiety and despair regarding the potential timing of onset and severity of the disease («*Despite the history of cancers in my family, it was a huge shock! I couldn't help thinking about the moment I could be diagnosed with cancer*», Beatriz, female, 35

years old); symptomatic participants, or with symptoms remission, were mainly worried about the descendants' risk levels, as their medical condition was being controlled («*My situation is under evaluation and I trust doctors, but what about my daughters and their future?*», José, male, 50 years old). Participants with positive results also reported feelings of guilt and self-stigmatisation:

> ***"****It is like being labelled "different" when I compare myself to others, despite knowing that it's not my fault…; I am concerned that the guilt I feel may turn against me, and I might end up being blamed by my kids, for instance"*. [José, male, 50 years]

## 1.4. DISCUSSION

This study reports the experiences of ten people regarding genetic counselling for hereditary cancers. Participants shared their perspectives on the different stages of the genetic counselling timeline, providing conceivable data regarding adjustment for hereditary disease risk.

Since the first consultation, where clients are informed about hereditary inheritance patterns implicated in family illnesses, genetic counselling is perceived as a complex and scientific procedure. After the family medical history is explored, clients are informed about their eligibility for genetic counselling and testing (together with other family members who are at potential risk); at this moment, the perceived genetic risk is experienced individually with considerable psycho-emotional tension, while family "threats" are considered.

The probabilistic nature of genetic outcomes (predictive and not prophetic, as stated by Werner-Lin, 2008) and the absence of indisputable benefits of knowing one's genetic risk create an uncertain context that surrounds the decision making process. Once the test results are known, the client is given the chance to know if he/she carries an identified pathogenic gene mutation. This psychosocial environment is permeated by acute uncertainty and ambivalence, given that the risk reduction measures available usually involve invasive procedures (*e.g.* prophylactic mastectomy or colectomy).

Participants stated that genetic counselling is a valuable step in an ongoing (chronic) process of genetic risk, especially because it allows them to acquire knowledge and education about genetics and genetic risk, which helps them gain a sense of control over health management. There is evidence (Hurley et al., 2006; Weil, 2005) that genetic counselling encompasses diverse practical and emotional challenges, confronting the client with significant uncertainty (concerning disease inheritance, timing of onset and severity of the disease, preventive and prophylactic procedures and its timings, personal and family future planning) and variable levels of psychological distress (since procedures are permeated by anxiety, worry and doubt). Consequently, participants associate coping with genetic illness with having a sense of control over one's health, which is mediated by

the degree of (un)predictability involved and by the threat of a genetic disease (enhanced by positive test results).

### 1.4.1. Recommendations

The following recommendations, arising from participants' reports, aim to contribute to improving the psychosocial focus on genetic counselling:

i) The genetics healthcare team should include a skilled professional in the area of psychosocial genetics to assess the clients' routine concerns and to provide them and their families' psychosocial support throughout the genetic counselling timeline.
ii) Psychosocial support should be made available since the pre-test counselling phase, including a follow-up session one month after receiving the test results, focusing on the following aspects: improving clients' and family members' psychosocial adjustment in the face of the emotional complex and uncertainty; and supporting decision-making;
iii) Providing educational written literature and a website especially designed to address medical information, psychosocial issues, and community resources; and creating a blog where people can share their doubts, worries and experiences.
iv) Facilitating psychosocial group support for clients/families, during the different phases of genetic counselling (pre-test, between test and results, and post-test).

Experiencing genetic counselling may cause significant distress, assuming variable dimensions according to the client's genetic illness time phase and its position throughout the genetic counselling timeline. Therefore, these recommendations call for collaborative work among different health care providers, in order to develop innovative interventions intended to address the holistic needs of those seeking help from genetic counselling services (Speice et al, 2002; McDaniel, 2005).

### 1.4.2. Limitations and research perspectives

Data should be deepened through further research using different methodologies, such as individual in-depth interviews or life stories. When exploring genetic counselling experience, it is important to address its impact on family dynamics, namely by analysing other family members' perspectives on how to best handle the uncertainty that surrounds genetic counselling. Further research should produce data regarding individual and family dynamics beyond the phases of genetic counselling, in order to provide an intervention process also based on their psychosocial needs. The implications of the present recommendations should also be addressed.

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## 2. FAMILIES´ EXPERIENCE OF ONCOGENETIC COUNSELLING: ACCOUNTS FROM A HETEROGENEOUS HEREDITARY CANCER POPULATION[26]

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**ABSTRACT**

This paper reports the results of semi-structured family interviews conducted with a purposive sample of nine families (comprising 50 individuals) involved in cancer genetic counselling at a Portuguese public hospital. Qualitative analysis resulted in thematic categories illustrating: i) how families go through cancer genetic counselling (eliciting risk awareness, the motivators, risk management, the psychosocial context of familial engagement in genetic counselling, and the familial pathways of cancer risk tracking); and ii) how families incorporate genetic risk into family life (strategies for family resilience, and the meanings and values that permeate the experience). Families have recognised the value of genetic counselling in enabling participants to take measures to confront disease risk; however, the experience was dominated by distressing feelings. A set of ethical-relational principles guided the experience. Familial experiences on genetic counselling and tracking of cancer susceptibility encompass a sense of trajectory that takes the form of an historical and intergenerational narrative process, linking past, present and possible futures. Such process implies an ongoing set of individual and interactional experiences taking place over time. Specific changes associated with the illness timeline and with individual and family developmental lifespan transitions are thus acknowledged. These results may help genetics healthcare practitioners understand how families perceive, respond to and accommodate cancer risk counselling, and thus illuminate family-oriented tenets for planning and practice.

**Keywords:** genetic counselling; family; hereditary cancer; cancer risk; grounded theory.

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## 2.1. INTRODUCTION

Genetic conditions, such as hereditary cancers, carry both health-related and psychosocial implications for the family system as a whole, even if only some family members are actively engaged in genetic medicine practices [1-5]. Families' views on genetic counselling are crucial, because when a condition is thought of as genetic, it is no longer a strictly individual matter. It is a family matter, as it involves family history, current life decisions, and potential family futures [6, 7].

Despite some exceptions [8-11], those studies which involve or are about families and genetic counselling issues have focused primarily on the proband and related specific relationships, not fully considering the familial environment and interactional milieu in the family system. Our study is not limited to relationships involving the proband, as we have decided to also include other family members. Considering the family as a unit will help to promote a better understanding of how families perceive, respond to and accommodate oncogenetic counselling in health management and into their lives.

### 2.1.1. Hereditary cancers

Rapid developments in cancer genetics knowledge and cancer prevention and early detection pose genetic testing for cancer pre-disposition as an increasingly common clinical practice. It is estimated that 5–10% of cancer cases are due to inherited mutations [12]. Hereditary non-polyposis colorectal cancer (HNPCC) and hereditary breast and ovarian cancer (HBOC) are the most prevalent types of hereditary cancer syndromes, both implying an increased risk of developing cancer. In mutation carriers the risk may increase to 80%, although variable penetrance remains uncertain [13]. The psychosocial implications of increased cancer susceptibility are well documented, frequently resulting in an experience of anxiety, fear, worry and doubt [14-17]. A negative genetic test result may also become burdensome, resulting in feeling responsible for the future care of relatives with the mutation, fractured relationships with mutation-positive family members, or survival guilt [18]. Cancer risk management for mutation carriers is complex and includes risk-reduction surgery options (mastectomy and/or oophorectomy for HBOC, colectomy for HNPCC, and gastrectomy in the case of hereditary diffuse gastric cancer, HDGC), intensified screening and surveillance (mammogram, gynaecological ultrasound, CA-125, colonoscopy, endoscopy), or chemoprevention (for HBOC) [13]. Some of these interventions are reported as invasive and are often complicated by life planning issues such as partnering, childbearing, career, or lifetime decisions, depending on the family's developmental stage [19, 20].

### 2.1.2. Mapping genetic cancer risk counselling as a family issue

The focus upon the family is intrinsic to the practice of genetic counselling. The counselled individual is located within biological and relational bonds, and familial memories, involving mutual obligations and ethical dilemmas as well [7]. When multiple genes are involved in the causation of disease, as with hereditary cancers, identifying the mutation responsible for the disorder in an affected relative is the common procedure to: first, know the specific mutation; and then develop a genetic test to identify it [12]. In fact, the familial input is present in cancer risk counselling from the very beginning. The first interview with the client mainly addresses family issues and is only in part about the client him/herself. During that interview the client's potentially inherited health risks are searched through the family history, which provides information for cancer risk assessment and DNA diagnosis (genetic testing). The drawing of the family's pedigree requires the clients to recall relevant information about the members of the last three generations of their family, such as date of birth and death, cause of death and the histories of illnesses for each member [20]. The client who does not have all the information is advised to inquire with his/her relatives in order to track down the missing information. Additionally, medical genetics practitioners typically rely on the proband to inform relatives about their potential at-risk status and predictive test availability [20, 21]. Subsequently, they also rely on other family members who come forward for testing, in a procedure that to some extent makes those members responsible for sharing information with additional family members.

Research has shown that responsibility for disclosing genetic risk information typically lies on family members [11, 22-26], influencing not only the uptake of genetic testing, but also the patient's adherence to prevention and early detection measures [27, 28]. Communication about genetic counselling or testing is most likely to occur among first-degree relatives or spouses and when relationships are defined as cohesive and without conflict [28]. Open communication and familial emotional support regarding hereditary cancer can function as protective buffers against distress [17]; significant others, as friends, tend to assume the role of family supporters [29]. Furthermore, inherited genetic risk demands ongoing psychosocial adaptation for those genetically at risk and their families [16]. Familial cancers commonly reveal multigenerational patterns of illness manifestation and can shape families' developmental processes, as beliefs and expectations about the individual and the family life cycle can be considerably affected. The interactional system between the individual, the family and the genetic illness timeline assume particular clinical significance. Transitional changes concerning the carrier status, symptoms onset or decision making regarding prophylactic interventions are psychologically challenging [30, 31]. In fact, ambiguities and uncertainties of threatened futures when considering genetic testing or confirmed mutation carrier status are clearly out of synch with those normative expectations which are

associated with several life cycle stages, often involving anticipatory loss of personal and family development goals [32, 33].

### 2.1.3. Genetic counselling and family systems perspective

Genetic counselling is indissolubly connected with family adaptation to inherited illnesses, assuming a key role in the incorporation and dissemination of genetic information through family members, in their health management, and in life planning issues. Research states that the family influences, and is influenced by, the response, the use, and the meaning ascribed to genetic risk [34, 35]. Attachment theory has been linked to genetic illnesses due to the recurrent and expected repetition of losses, leading to the reconfiguration of family roles, and often hindering reliable secure attachment experiences across generations [36]. The level of emotional differentiation from parents has also been reported as crucial in shaping psychological adjustment within families [37]. Family cohesion and adaptability are major characteristics of family functioning [17, 38]; cohesion represents emotional bonding; adaptability corresponds to the ability to change family structure, rules and role relationships under specific needs or demands. A balanced level of these characteristics has shown to be beneficial for familial adjustment to stress [38]. Families exposed to inherited risk tend to define their identity based almost exclusively on that experience; this reorganisation around the ambiguous territory of at-risk status and the need to develop coping efforts under emotionally charged contexts can lead to rigidification of family functioning and development, giving the family the identity of a *genetically ill* family [8, 39].

A family systems perspective offers a potentially useful framework for understanding family issues in the scope of genetics [4, 5, 40, 41]. A conceptual framework addressing the set of psychosocial issues faced by families with genetic conditions was provided by Street and Soldan [40], based on Rolland's previous work on chronic illnesses [42]. These authors have proposed an expansion of the time phases (course) of the illness, because of its insufficiency to account the time before the illness onset inherent to some genetic illnesses – *the pre-illness phase*; this is especially relevant since in many cases mutation carriers live pre-symptomatically for a considerable amount of time, before visible symptoms of the disease become noticeable. Subsequently, Rolland [43] and Rolland and Williams [4] have clarified the timeline of genetic illnesses, distinguishing its non-symptomatic and post-clinical onset phases, each with sub-phases and related psychosocial demands. Rolland [32] has also conceptualised the psychosocial interface between the time phases of genetic conditions through the individual and family life cycle, suggesting a way for health practitioners to consider it longitudinally as an ongoing process with transition points and changing demands. The influence of the anticipatory loss was also acknowledged; living with uncertainty due to possible, probable or inevitable future loss is challenging for individuals and their families; balancing the involvement of a person who may develop a life-threatening illness, or assuming

caregiving responsibilities for an already ill relative, while maintaining a flexible family functioning and considering future life planning, are certainly a serious endeavour [32, 42, 44].

As genetics healthcare practitioners become increasingly educated about predictive medicine and pre-symptomatic genetic testing, it is relevant to include family-oriented data into their understandings, namely regarding the way families manage their experience of genetic knowledge, testing and counselling, and how it influences family identity. Addressing the family environment and dynamics has been reported as crucial for research [45], and for assessing psychosocial needs [46, 47]. This exploratory, qualitative study aims to explore the families' experiences of cancer genetic counselling, through a familial perspective. Our results contribute to a growing body of work on the effects of genetic counselling in families, perceived risk and family history.

## 2.2. METHODS

Grounded theory is described as both a systematic and rigorous method, allowing participants to be the experts in describing their own experience [48, 49]. It is often used to explore topics where little is known to guide research or practice [49]. Grounded theory is designed to explore how people understand their circumstances and how they describe and define a given specific situation, as well as the manner how such understanding is related to action [49, 50]. This study used a grounded theory design because this approach enhances the familial perspective on how participants experienced a given procedure (genetic counselling) and the related psychosocial environment.

### 2.2.1. Recruitment

Participants were recruited through purposive sampling from the Centre of Medical Genetics and Human Reproduction of the University Hospital of Coimbra, Portugal. Ethics approval was granted. The eligibility criteria consisted of: consultands over 18 years of age, who had previously undergone genetic counselling, from families with at least three known mutation carriers, regardless of whether they had previously been diagnosed with cancer. Eligible individuals transmitted research study information to family members whom they might considered relevant to participate in the interview. Researchers were dependant on index patients to assist with the recruitment of other family members; researchers were cautious not to interfere with patient and familial autonomy, or steer towards contacting any other family members. The recruitment process involved searching through the service database to identify eligible subjects; 12 subjects were then selected. These subjects were sent a letter briefly outlining the study and inviting them to take part in a family interview, while mentioning a subsequent phone call to address availability to participate in this study. A phone call was made two weeks later aiming to explore the feasibility of a family interview, namely in terms of contacting other family members to participate in the family

interview (participants were told that any family member, whether or not related by bonds of consanguinity, whether or not they had a genetic testing done, and whether or not they were mutation carriers, was eligible to participate in the family interview, including significant others). Two weeks later, a second phone call was made to those individuals who had agreed to participate in order to assess if they still agreed to cooperate. At this point, 2 of the individuals who were eligible declined to participate; due to ethical reasons, motives were not explored. Those who agreed to participate were asked permission to be sent additional recruitment letters which would then be distributed to other family members; the letters were sent and a subsequent phone call was made to schedule the interview; one consultand dropped out after the initial agreement.

The recruitment method used cascade sampling, following similar practical paths commonly used by families to disseminate information about genetics. This is an innovative method to assemble research participants, since it largely relies on how families organise themselves to enhance communication between all of their members, and for that reason, it is dependent on the structural characteristics and relational peculiarities of each family. Therefore, geographic proximity and relational closeness between family members arguably assume a pivotal role for mobilising participants to the interview.

### 2.2.2. Participants

The sample comprised 9 families (50 participants) (Table 2.1.): 5 were affected by HNPCC mutations, 3 by HBOC mutations, and 1 by a HDGC mutation. Participants consisted of individuals from both nuclear and extended families, mainly comprising first-degree relatives (parents, offspring and siblings) (88%); other family members included in our sample were spouses.

**Table 2.1. Participants (*n* = 50) demographics, illness characteristics and family history**

| **Characteristic** | **Family 1** (*n*= 6) | **Family 2** (*n*= 5) | **Family 3** (*n*= 5) | **Family 4** (*n*= 5) | **Family 5** (*n*= 6) | **Family 6** (*n*= 5) | **Family 7** (*n*=5) | **Family 8** (*n*=7) | **Family 9** (*n*=6) |
|---|---|---|---|---|---|---|---|---|---|
| Mean age | 55,0 (35-73) | 55,2 (33-78) | 31,1 (15-41) | 36,0 (19-50) | 43,0 (22-62) | 39,0 (31-58) | 55,0 (19-78) | 42,0 (24-57) | 47,3 (26-76) |
| Females | 4 | 5 | 4 | 2 | 1 | 3 | 4 | 2 | 4 |
| Mean years of education | 10,3 | 8,0 | 8,4 | 10,0 | 8,6 | 11,4 | 7,8 | 5,5 | 8,6 |
| Generations represented | 2 | 3 | 2 | 2 | 2 | 2 | 3 | 2 | 3 |
| Biologically related family members | 5 | 5 | 4 | 4 | 6 | 4 | 5 | 6 | 5 |
| At-risk individuals | 4 | 5 | 3 | 3 | 5 | 3 | 4 | 5 | 4 |
| Former cancer patients | 2 | 3 | 1 | 1 | 1 | 1 | 2 | 2 | 2 |
| Cancer-related deaths | 3 | 3 | 3 | 2 | 3 | 1 | 2 | 8 | 3 |
| Genetic testing uptake | 4 | 5 | 3 | 3 | 5 | 4 | 4 | 6 | 4 |

### 2.2.3. Interview guide

For this qualitative study, a semi-structured interview guide was developed (Table 2.2.). As background to the study, we relied on literature about the effects of chronic illness on: family functioning [39, 42], familial aspects of inherited risk [4, 5, 43, 51], and prior research findings [52].

**Table 2.2. Interview guide**

| Topics | Questions |
|---|---|
| Risk awareness and motivators for genetic counselling | *Tell me about when you began to think that a hereditary illness might exist in your family?* |
| | *What have made you decide to get genetic testing? Why you had accepted to do it?* |
| Mapping the family experience | *As a family, how do you characterize your experience with genetic counselling? What were its positive and negative impacts, and the most difficult/easiest moments?* |
| | *What have you done to cope with genetic counselling and, after results were known, to the heightened risk?* |
| Meanings, values and family identity | *What meanings genetic counselling brings to you? What does it mean for you to belong to a family in which it runs a hereditary illness?* |
| | *How the experience of genetic counselling and the increased genetic risk does change the way you feel as a family?* |
| | *What can make genetic counselling a positive thing for families?* |

### 2.2.4. Data collection

#### *2.2.4.1. Interviews*

Nine families comprising 50 individuals were interviewed as a family unit in their homes for approximately 1.5 hour; all interviews were taped with the interviewees' consent and later transcribed. Interviews aimed to explore as fully as possible the familial experience of the process of genetic counselling and the meaning, impacts and management of the genetic condition. A genogram [53, 54] was initially created to collect demographical data and illness-related features, followed by an initial open question inviting participants to tell the story of how their referral to the genetics clinic came about. From there a series of exploratory questions was used, mixing open-ended questions with more focused questions (Table 2), while probing important topics as they arose; the interview schedule was dynamic and questions used in later interviews were developed in the light of emergent findings. The semi-structured nature of the interview allowed families an opportunity to "tell their story"; circular questioning was used [55]. The interviewer adopted an active collaborative role in the process, engaging with participants in ways that influenced the shape and flow of the conversations, while clarifying and summarising their accounts. Theoretical

memos and case summaries were written down from each interview to highlight the most relevant aspects and contextual observations, as well as to record ideas about the emerging data [50].

#### *2.2.4.2. Post-interview questionnaire*

Three weeks after the interview took place, participants received an individual questionnaire, with their consent, in order to address the personal relevance of the family interview. Questionnaires were sent to participants by postal mail, along with a postage-paid return envelope. In this questionnaire participants were asked to make a brief statement concerning how they felt about being interviewed together as a family; they could also include any additional comments they wished to make as helpful ideas for researchers when meeting with other families. These post-interview questionnaires were adapted from Sobel and Cowan [9], and were used to provide a reliability check on data analysis and to give feedback on the emotional difficulties posed by the interview.

### 2.2.5. Data analysis

Data continued to be collected until it reached a point of redundancy [56]. All interviews were transcribed and submitted to content analysis. Transcripts were reviewed and read several times for interpretation; open coding was used to illustrate the specific features of the data deriving from recurring ideas and themes; memo writing [50] was used to analyse data and the developing codes, and for conceptual clarification during the research process. All data were coded by the first author and an independent researcher to enhance reliability [56]. The coded transcripts were compared for consistency, and then, one of the researchers (first author) continued the analysis based on a process of successive coding refinement by repeatedly reading the transcripts and grouping similar ideas into themes, aiming to develop emergent content categories. Categorisation titles were given to similar themes and contents.

## 2.3. RESULTS

Two major themes were identified in relation to the families' experiences of cancer genetic counselling, each comprising subthemes: i) going through genetic counselling (eliciting the risk awareness and the at-risk family status; engaging in genetic counselling; motivators, risk management, and emotional context; tracking risk: familial pathways; and ii) incorporating genetic risk in family life (strategies for family resilience; orienting values and meanings) (Table 2.3.). The first is a thematic category, encompassing salient features upon which families drew their cancer risk and genetic counselling experiences; the latter represents a cross-cutting, meta category which is embedded in the core of the families' accounts.

Themes were not mutually exclusive and represent fluid rather than discrete and sequential categories. Each major theme is presented along with quotes from participants to illustrate related key points. Quotations are followed by the participant's name and by a code notation indicating their sex (M/F) and the family they represent (number, see Table 2.1.). All participants' names have been changed to protect their anonymity. The initials "AM" refer to the interviewer (first author). Content in square brackets is used to add intelligibility to the participant's quote. Ellipsis dots are used to indicate a suppression of the participant's quote, aiming to highlight pertinent discourse.

**Table 2.3. Major themes and subthemes**

| **Going through genetic counselling** | | |
|---|---|---|
| Eliciting risk awareness and the at-risk status | Engaging genetic counselling: motivators, risk management and emotional context | Tracking risk: familial pathways |
| **Incorporating genetic risk in family life** | | |
| Strategies for family resilience | | |
| Orienting meanings and values | | |

### 2.3.1. Going through genetic counselling

#### *2.3.1.1. Eliciting risk awareness and the at-risk status*

Family health histories permeated by several cancer-related events, such as diagnosis, treatments, contacts with the healthcare system, or deaths, seem to have propelled the formation of risk awareness in the family context. A link between seeing close relatives being diagnosed with cancer and the perception of familial illness susceptibility was clearly evidenced by participants of all families:

> "*When my brother became sick I thought that something was running in our family, because the same thing had happened with my sister, who had died a couple of years before… I've seen two uncles on my father's side dying of cancer (…) At the beginning, you try not to think about it, but then, as time goes by, with the hospital, the treatments and everything, it's just too much to be a coincidence*." (Dinis, M3)

Most of the families provided detailed accounts of their cancer-related journeys. These narratives were rich in stories of sacrifice, involving the recapitulation of important dates, critical moments, and successful or negligent contacts with the healthcare system. An intergenerational path of risk awareness, due to the high prevalence of cancers affecting several family members, emerged in families with a "stronger" history of cancer:

"*When my grandmother died, I remember my father and my uncles saying that they were all gone* [the older generation]*… as I grew up, all relatives on my father's side either became ill or died*!" (Elias, M8)

AM: What things or stories can you recall?

"*Nothing much… I remember visiting my uncles at the hospital or at their homes; years later, I saw my cousins doing the same and visiting my father… it's like a cycle."* (Elias, M8)

In two HBOC families, the younger women stated the age when their mothers were diagnosed with cancer as something that shaped their expectations of genetic counselling:

"*When my mother was my age, she already had cancer; she was younger than me when she was diagnosed with cancer!*" (Maria, F2)

Participants shared their own explanation for the heightened risk in their families. Lay understandings of illness causation were mentioned, ranging from attributions of "fate" and "bad luck" to rough explanations of heredity, or health behaviour, such as dietary habits:

"*It's in our blood, and it passes on from one person to another…*" (Paulo, M6)

"*It's pretty obvious to me that this* [cancer risk] *came from my father's side; something happened back then, probably due to dietary food intake habits*" (Luís, M4)

All of the HBOC families interviewed evidenced a gendered awareness regarding their at-risk status. One HNPCC family mentioned the disease as a "man's thing", even if the genogram had clearly elicited affected females in the extended family:

"*I think this is a man's thing (…) only my son got it* [the deleterious genetic mutation], *the girls didn't get it!*" (Susana,,F8)

"*I talked to my sister and then to my cousin, because, as we're women, we feel we must do something; looking back* [through our family's medical history], *you cannot avoid thinking that women have been particularly exposed to breast cancer*" (Joana, F1)

Some families reported open and frequent talks about cancer risk, while others were reluctant to engage in such conversations:

"*You know, it's something that you think about but you don't actually talk about it. You just do what it takes and keep moving on; we do talk about it when there's a consultation or a colonoscopy, but you just cannot live constantly thinking about it, because this is something we've been carrying for a long time*." (Josefina, F5)

#### *2.3.1.2. Engaging in genetic counselling: motivators, risk management, and emotional context*

All participant families came for genetic counselling through a physician referral. Consideration for genetic testing was primarily viewed as a chance to be informed and to act for prevention:

> "*I wanted to know and to understand where it came from; the more you know, the more you're aware of the things you can do; you can't just stay seated and wait for the rock to roll over your head!*" (Dinis, M3)

The prospect of extending the service to other family members, especially descendants, was emphasised as another major motivator for participants' involvement in genetic testing; families valued the possibility to obtain information about how to be proactive with their own health and to improve their well-being while preparing for the future. Some families reported that the older members had encouraged the younger generation to engage in genetic counselling:

> "*I made the test because I wanted to protect my children… I wanted them* [daughters] *to know, so they could be able to get something done as soon as possible*" (Serafim, M8)
>
> AM: What happened then?
>
> "*My dad was worried because he was dealing with a relapse when the positive result was known. It was disturbing for us because I was about to get married and I just didn't want to hear about all that …and after a year or so I took the test and it was negative*" (Margarida, F8)

Finding the origins and routes of the condition was also a motivator for genetic testing:

> "*I guess I wanted to know where it came from; it's important to look back and see where it started*" (Joaquim, M4)

Knowing specific information about their risk status seemed to lead to further possibilities of action:

> "*I knew that I was probably at-risk; I was expecting a positive result, but a confirmation was important for planning my* [preventive] *surgery. I needed to be sure. Due to our family history of cancer, I felt I had to do something!*" (Monica, F9)

Access to medical expertise and recent technological advances gave some participants a sense of empowerment:

> *"I was amazed by the chance to be seen by a specialist doctor, which was a really good opportunity (…) technology gives you the possibility to know more, and almost immediately (…) if the doctor advises you to do something, then you should take the advice!*" (Maria, F2)
>
> "*I went to genetic counselling to be able to understand what was happening and to learn about prevention; I wanted to know what I could do to reduce risks, to engage in tight surveillance, and to gain control*" (Gabriela, F7)

Implications of genetic counselling were mostly felt in three major domains: i) inherent demands of the genetic counselling process, ii) its emotional impact, and iii) in family developmental transitions. Dulce and Filipe were planning to get married; as they were third-degree cousins (this

is relatively common in some rural areas in Portugal), when Dulce found out about their carrier status for HNPCC, Filipe also got tested and he turned out to be a carrier as well. When Dulce got pregnant, they both underwent genetic counselling, without prenatal genetic diagnosis:

> "*Dulce told me that she was a damaged doll because of that* [carrying the deleterious gene mutation]. *(…) when I found out that the gene was in me as well, we both got scared, because we wanted to have many children …*" (Filipe, F7)
>
> "*It was a huge shock to find out that he also had it* [the faulty gene] *(…) As we were trying to find out how we would cope with all that, I got unexpectedly pregnant; we received* [pre-natal] *genetic counselling, but that was so confusing… the options to reduce the risks for the baby were too complicated, involving some kind of gene therapy in the Netherlands…* [pre-implantatory diagnosis]" (Dulce, F7)

Assimilating new information about heredity and genetics was the first requisite families recalled as being needed, despite difficulties in fully understanding the information:

> "*Some things were hard to understand, but I think I could grasp the essential*" (Joaquim, M4)
>
> AM: Which things were hard to understand?
>
> "*Things were too fast for me, I think I needed more time to digest all the information*" (Luís, M4)

As genetic test results were known, and (variable) risk was confirmed, several families reported decision-making for risk reducing options, which were perceived as a valuable help, especially by those who had already undergone prophylactic interventions:

> "*I was really afraid of it!* [prophylactic total gastrectomy – preventive surgery involving complete removal of the stomach] *But I was very well treated and as everyone knew about it, that wasn't a complete surprise, and I think that really helped me. I had to do it, otherwise I'd be at high risk and there would be a higher chance of getting* it [gastric cancer]. *So, I never had doubts about the final decision!*" (Diana, F3)

Mutation carriers also reflected hope in preventing the disease through increased surveillance measures, while delaying prophylactic interventions; life planning issues were also mentioned:

> "*I feel more protected, because I have my mammograms and ultrasounds done, and I know what I have to do to prevent something from happening!*" (Maria, F2)
>
> AM: Have you considered any prophylactic measures with your doctor?
>
> "*Yes, but only in the future; right now, I am doing yearly screenings, because I still want to have kids*" (Maria, F2)

However, it is a stress-laden process that carries ambivalence, and a strong sense of vulnerability:

> "*I remember feeling constantly worried about having a recurrence, and I also worried about my daughters' future… I've been through a lot, but there's always this fear, this kind of shade upon me and my family…and it comes and goes… right now, I'm trying not to think about it and move on*" (Irene, F2)

Families dealt with successive stressful demands due to the cascade of events imposed by risk management. Sara spoke about her experience as a caregiver, vividly illustrating the family context as a permanent state of dealing with uncertainty:

> "*Seeing my husband dying was unimaginable, but all those months* [waiting for daughter's test results] *were a huge fight for me, because I had to stay positive, to support and encourage my daughter to do the surgery (…). So, now we just want to move on, but soon we'll have to think about Luísa and Vera* [her nephew and her youngest daughter, aged 15 and 12, respectively], *and deal with it all over again*" (Sara, F3)

Feelings of fear, shock, worry and angst were described as permeating the process of cancer genetic counselling, even when participants reported that a positive test result was already "expected"; waiting for test results and the confirmation of a genetic diagnosis were particularly stated as emotionally difficult moments:

> "*I remember that when I got the result, I couldn't do or think about anything. I called a friend to pick me up at the hospital and take me home. I was devastated!*" (Dulce, F6)

Participants, especially older ones, showed their struggle with guilt, something which was also reported concerning relatives who were not present. Manuela (non-affected mutation carrier) revealed:

> "*Every single day I ask God why…it's something I've been carrying with me since it all* [mutation screening] *started*" (Manuela, F9)
>
> "*I used to feel a lot of angst…in a way, I felt I was responsible for my daughter's disease… I can't really express how I feel right now*" (Adelaide, F6)

A small number of non-carriers integrated the families' interviews; Elias described how he felt for being one of the few in his family with such genetic status:

> "*It's weird because my brother got it and my father got it as well…there are some more cousins like me* [non-carrier], *but as I got lucky on this I also feel bad for them; I don't feel pity for them, they're doing fine and I know they're happy for me. I cannot blame myself, but sometimes I feel like it isn't real… I feel that I don't deserve it*" (Elias, M8)

#### *2.3.1.3. Tracking risk: familial pathways*

Communicating with relatives emerged as a topical aspect during families' involvement in genetic counselling, both for transmitting information about potential increased risk and for gathering

information on the family's health history. Serafim recalled his experience of asking a member of the older generation for specific information to help establishing the family pedigree:

> "*I was the first to go there* [to genetic counselling] *and I brought a lot of homework! I was unsure of a few specific details related to my cousins, so I talked to an aunt of mine who knew about the dates and the time when they all got sick*" (Serafim, M8)

All families had, to some extent, disseminated genetic risk information to potentially at-risk relatives, doing it in an open way within the nuclear family and first-degree relatives. Informing extended family members was more problematic for some families because of emotional distance, rifts or reluctance in bothering them:

> "*We are a family of 7 siblings and some of us don't get along very well (…). I left those* [informative] *sheets they gave us at the hospital at my mother's, because I know they'll go there. My wife talked to my sisters-in-law, but I don't know if they have already been there*" (Joaquim, M4)
>
> "*I didn't want to worry them* [uncles and cousins] *about this… besides, my cousin was exactly the same age as my uncle was when he died 10 years ago… but anyway I ended up telling him about all this and he got pretty scared… I don't know if he has already taken the* [genetic] *test*" (Filipe, M6)

In one case, the family disclosed genetic risk information to younger members. Family 3 openly shared their expectations regarding prophylactic measures with the adolescent Julia, while protecting the other two younger children (absent at the interview). Julia showed herself determined to undergo testing after she turned 16, the age her cousin Diana got tested, despite being anxious about the gastrectomy, the most likely scenario in case she was a carrier: "I know that if the test is positive I wanna do the surgery…but what if things get worse?".

The index-patient of each nuclear family (the first family member to undergo genetic testing) typically assumed a two-folded role of being the messenger and the gatherer; more than one relative was often engaged in disseminating information to other family members, which tended to occur in an intergenerational manner:

> "*I was the oldest in the family to be diagnosed with cancer, so they tested me straight away, and then I talked to my daughters and my sister; and my daughters talked to my nephew*" (Maria, F1)
>
> "*I've got a large family… I talked with many of my cousins, and of course with my daughters; and when they got tested, along with my nephews, I guess they did the same; you just have to spread the word*" (Serafim, M8)

Women (mostly spouses of former cancer patients) seem to maintain and facilitate communication among those relatives with distanced or troubled relationships, while assuming themselves as a source of emotional support:

> *"I think it's easier for me and my sisters-in-law* [to talk about cancer risk and genetic testing uptake with their husbands] *(…) some of them were sick a few years ago and they just don't like talking about this issue all the time; it's also a way of supporting each other*" (Alda, F4)

Blockers to the exchange of information about cancer risk were mentioned in two cases:

> "*I've always told everyone about this* [heightened cancer risk], *but my father and my older brother were very difficult to convince… They think we should not inform the kids about this, and they don't allow us to talk to them*" (Dinis, M3)

Several families suggested that the medical team should have a proactive role towards potentially at-risk relatives (particularly from the extended family) in order to improve genetic testing uptake: this approach was viewed as a way of adding credibility and accuracy to what is shared among relatives, and it could alleviate the burden that somehow families feel in this process:

> "*Doctors should talk to them* [potentially at-risk relatives] *because in many cases they don't listen to us (…) they live in distant places, and we only see each other from time to time… besides, we're not doctors, we don't know exactly what or how to say it!*" (Armando, M8)

### 2.3.2. Incorporating genetic risk in family life

#### *2.3.2.1. Strategies for family resilience*

Increased genetic risk for cancer poses ongoing efforts for adaptation. Closeness and strengthened family union were reported both as a consequence of cancer risk and as a crucial trend either to provide emotional support and encouragement or to monitor adherence to medical surveillance recommendations:

> "*We have always been a very close and united family; this* [knowing her increased susceptibility for familial cancer] *has brought us even closer, we call each other when someone goes to a consultation, we often get together and talk about everything, it sort of helps to avoid becoming stuck and hopeless*" (Madalena, F2)

Attention to medical recommendations assumes pivotal significance for the majority of the families; some consistently mentioned healthcare practitioners as influential in supporting decision-making, namely in the process of transmitting information about risk to kin:

"*They were very clear, my cousins should be warned about it the sooner the better! And that's what I did. Who am I to ignore such a thing?! A good doctor needs good patients!*" (Serafim, M8)

"*I believe in doctors… it's how I tell them* [4 sons]*: the only thing we can do is to do everything they* [health care practitioners] *tell us, and then whatever may happen, we cannot control it*" (Rui, M5)

Religious faith and the adoption of an optimistic perspective were also reported:

"*(…) we try to keep ourselves optimistic about all this and carry on with our lives, without whining*" (João, F4)

"*We aren't very religious, but I think faith gave us some comfort in the worst moments… In such moments we hold ourselves to what gives us hope*" (Cândida, F9)

***2.3.2.2. Orienting meanings and values***

Families' interviews evidenced that participating in genetic counselling is beneficial and empowering; genetic test uptake was viewed as useful for disease prevention and health management:

"*I feel more protected because I know that I'm being studied, it kind of gives me more confidence; we look into the future with a more positive feeling*" (Maria, F2)

Genetic counselling and risk reducing options were viewed with a pragmatic sense to pursuit family planning:

"*It was likely that I would get cancer in a few years' time …We cannot just let things happen, we must use the resources we have (…) I realised that my life was worth more than my breasts; I got pregnant after the surgery, and now we have a baby…*" (Mónica, F9)

Metaphors illustrating being part of a high risk cancer family came linked with optimistic perspectives regarding a "normal" life:

"*It's like when cars are produced with a faulty part which is only detected later on, and the manufacturer advises you to take the car, so that the damaged part can be fixed… and then you can drive your car again!*" (Vitor, F7)

Despite the optimistic gaze, families consistently invoked genetic counselling as reflecting a stressful experience:

AM: Could you tell me what it means to be part of a high risk cancer family?

"*It's like walking on the edge of a sword… if you slip…; we try to carry on and we know that we have to keep an eye open, and stay alert*" (Sara, F3)

An altruistic relational-moral ethics permeates families' engagement in genetic counselling and related activities; this was particularly evidenced by the desire to give other family members the

opportunity to benefit from genetic testing (and from the same health management opportunities they had), especially children:

> "*Now we have to give our children all that the others couldn't benefit from; they're next, it's our duty*" (Sara, F3)

The desire to enable children to prevent disease in the future reflects a sense of hope in further technological advances for risk management options:

> "*To protect our children we must first know what happened with us; and as science evolves, it is most likely that when kids reach the age where they start thinking about it* [possible cancer risk], *things will be so advanced that they'll get much more prevention and healthcare*" (Dulce, F6)

Most families mentioned a sense of moral duty that has clearly shaped the process of informing potentially at-risk kin about attending genetic counselling:

> "*I had to do it; I couldn't just leave them in their ignorance!*" (Joaquim, F4)
>
> "*Our family needs to be studied because we share the same gene and everybody has to be warned about it; by doing so* [warn kin] *I get a clear conscience*" (Dulce, F6)

### 2.3.3. Post-interview questionnaires

Of the 9 families, 6 (65%) returned the questionnaire; of 50 participants, 30 responded (60%). 27 respondents felt that it was a good idea to be interviewed as a family; 14 respondents felt that being interviewed together with other family members was useful, 6 felt that it was intrusive/painful, and 10 reported that the interview was emotionally neutral. 18 participants made comments: 2 indicating that the family interview triggered difficult feelings about the past; 3 stating the benefits of sharing points of view with other family members; 5 reporting that the interview provided a means for them to hear about the views of others; 2 reporting relief; 1 pointing the joint interview as beneficial because of busy agendas; 3 describing the interview as a good opportunity to strengthen family bonds; and 2 stating that the interview was an opportunity to get better informed.

## 2.4. DISCUSSION

While tackling the familial experience of genetic counselling for hereditary cancers and its heightened risk context, this study offers important insights into prominent issues regarding adaptation in high-risk cancer families: first, showing the influence of family medical history on how families begin to acknowledge inherited cancer risk; second, showing the way they perceive and respond to the cancer risk counselling process; third, highlighting the specific tenets of intra-familial communication about cancer risk; fourth, displaying how at-risk status is integrated into

the familial milieu; fifth, revealing the meanings and values families ascribe to their genetic counselling experience.

This exploratory study adds to the literature in that it looks at how cancer risk counselling is experienced through the perspective of the families. While most studies addressing familial experiences rely in individualistic descriptions from one or more family members, this study employed family interviews to generate joint accounts on the families' experience. Its interdependent and collaborative nature enacts dynamic narratives expressing the contingencies of relationships, family identity, and the ways families create meanings in an interactional sense-making setting [57]. This approach is consistent with the need to acknowledge family interrelationships in the genetic counselling process, aiming to include family-oriented data as an intrinsic part of the provision of cancer genetic counselling [6, 41, 43].

In this study, 88% of participants were biological relatives and 12% were spouses, evidencing that genetic risk is a family affair which is primarily constructed and shared under intimate and close relationships. Only 4% of participants were non-carriers, and 72% were healthy mutation carriers; 30% were former cancer patients. In the light of these results, one might hypothesise that families perceived the family interview as directed to those who were primarily linked by the illness or at-risk status, under the gaze of the "cancer bond" [10]. On average, each interviewed family represented 3 households (27/9), including 2 to 3 generations. From our data we may speculate that participant families tended to live geographically close, but such data certainly indicates high levels of family cohesion.

### 2.4.1. Family history and formation of risk awareness

Findings elucidate the pivotal role of the cumulative effect of cancer-related events (diagnosis of cancer, cancer recurrence, and deaths) in giving rise to increased concern about cancer risk [10]. Each person starts by comparing the history of cancer-affected relatives with their own history and the history of their children; for example, reaching the age of the affected mothers seemed to add a supplementary feeling of vulnerability to HBOC [58]. Therefore, risk awareness is not merely elicited by the hereditary nature of the genetic mutation; there is an expectation or anticipation of "having the disease" because it is embedded in family history. Participants mentioned the need to understand what is happening to their family (why are so many people affected), finding out explanations on behavioural determinants such as diet, uncontrollable factors (fate or misfortune), or gendered considerations. Such lay understandings about genetic illness have been described previously [59, 60], whereas McAllister [61] advanced the term "personal theory of inheritance" to describe the individual's interpretation of his/her family history in HNPCC families.

Illness narratives about 'the thing that runs in the family' (the name of the disease is hardly ever mentioned), replete with stories of suffering, help to foster the importance of the intergenerational legacies in the families' identity. Sex-linked constructions, for example, may perhaps reflect a gendered behaviour derived from the fact that a cancer diagnosis is perceived as a crisis for the family system, with the potential to change the family's structural and interactional patterns; this poses an active role for women in elaborating illness causation explanations [61] and responsibility in exchanging information about risk with others [62], since women are described as the family health gatekeepers [2]. These pervasive accounts ultimately confirm the familial path towards cancer as a threatening future; while this sense of vulnerability achieves confirmation through a positive genetic test result, participants rely on their family narratives to interpret and create meaning for this vulnerability.

### 2.4.2. On cancer genetic counselling: motivators, risk management and emotional context

The need to cope with this kind of family "ghost" leads to different protective behaviours: the majority talks about it; the others avoid the subject. Yet, family history *per se* did not emerge as sufficient to lead them to seek medical advice. Key motivators for engaging in genetic counselling were: to obtain information conducive to act for illness prevention and early detection (through surveillance measures for early detection and prophylactic risk reducing interventions); and to enable relatives (especially descendants) to benefit from genetic counselling. Other factors emerged as relevant: older members were reported as encouragers for the younger generation in seeking testing and screening behaviours, which is consistent with previous findings [63]; and to make sense of the origins of the disease in the family. Therefore, engagement in genetic counselling entwines individual and interpersonal motivators, led by the influence of older generations, a commitment to monitor risk appropriately and to act to preserve other relatives' health, and to know the familial routes of the risk in the family.

Even though genetic counselling was perceived as empowering, since it adds further possibilities of acting in risk management (notably 'under the radar' of cutting-edge technological advances), in practice it was experienced as problematic for some families. Main problems rested in understanding genetic information, corroborating data from other studies indicating the difficulties clients experience in understanding probabilistic risk information [64].

Families also reported a considerable emotional burden. A cascade of successive stress-laden events enfolded an emotional complex led by vulnerability, angst, fear, guilt and uncertainty, permeating various key points of the genetic counselling timeline. For some families these events were described as chronic, ongoing stressors, requiring psychosocial demands for adjustment, due to the temporal perspective dominated by cancer risk management decisions and procedures.

Developmental strains emerged, namely through juxtapositions between individual and family life cycle transitions and the genetic counselling timeline. Young adults reported issues concerning family planning, such as marriage or childbearing, emphasising difficulties to achieve developmental goals and anticipating losses related to expectations about the future that genetic illness thwarts [30, 33, 65].

### 2.4.3. How does cancer risk information reach family members?

Participants naturally mentioned intrafamilial communication as a crucial feature in the process of tracking cancer risk within the family. Despite featuring as a motivator for families' involvement in genetic testing, exchanging information about cancer risk between relatives may also reflect the pragmatic focus derived from the cancer risk consultation, since encouragement for dissemination within potentially at-risk family members is a common practice for the genetic healthcare team. Additionally, participants also related that they tended to rely on medical advice to feel more protected. The following familial roles regarding the passing on of genetic risk information emerged: disseminators were also information gatherers; disseminators tended to have a personal history of cancer and were typically the proband (the first person in the family to undergo testing); blockers (absent at the interviews) to the transmission of information were also mentioned; older relatives were the "keepers" of families' health history, described as valuable resources and as providers of privileged health information, namely for family risk assessment. These results were generally consistent with literature [63, 66]. Families tended to mention that communication about genetic risk occurred openly within the nuclear family and among first degree relatives, whereas problems arose in communicating with extended family members (even though first degree relatives are here included) due to emotional distance, conflicts or the wish not to bother or scare them [67, 68]. Women (mainly spouses of former cancer patients) were active in helping to disseminate information in the latter cases, commonly mediating and facilitating these contacts, adding support to their role as "kin keepers" in the family [66].

Some families mentioned difficulties in informing relatives about genetic risk, pointing out ways to get help in this endeavour. Suggestions highlighted the ambivalent context this process attains: even though they feel committed to transmitting genetic information due to the existence of a moral bond to their kin, and despite being devoted to follow medical advice, families also feel burdened by the lack of professional guidance or support concerning this matter. Ethical debate on this issue enfolds three aspects: the clinicians' moral duty to warn clients, the right clients have to keep their medical information private, and the right other family members have (not) to know about their genetic risk [69-71]. Research on the intrafamilial pathways of communicating genetic information to relatives has shown that this process is influenced by family dynamics, family history, personal

understanding and ability to explain genetic concepts, and family members' interest and ability to understand the information [23, 72-74].

### 2.4.4. Coping with cancer risk: family resilience

Attention to medical recommendations assumed crucial significance for most of the families. One might hypothesise that engagement in medical advice has been part of the routine concerns of these families, constituting a way to validate their experience of suffering and to create meaning in order to face a very disturbing experience. In addition, relying on doctors promotes feelings of security and hope, especially because it leads to prevention. Relying on medical guidance emerges as a coping strategy for participants, since it leads to feelings of empowerment and agency, not only over one's health management, but also to enable extended kin (especially descendants) to be protected as well.

Families follow coping efforts to alleviate themselves emotionally, serving also as a means to integrate the at-risk status in their lives more functionally, and thus enhance a sense of mastery and resilience. Optimistic efforts to envisage the future appear as a straightforward aspect for managing the ambiguous territory of genetically-linked conditions. Augmented emotional proximity and union reflects high levels of family cohesion, which is coherent with the conceptualisation that poses chronic illnesses (such as hereditary cancers) as a centripetal effect on the at-risk family system, as a way to channel family members through unfamiliar and feared territories, helping to foster a sense of control that helps allay uncertainty [42]. Spirituality and religious faith is a good example that expresses the importance of sustained hope over anticipated loss of future plans and hopes, along with culturally-sensitive ways of constructing meaning. The notion of illusion of control is mentioned by psychosocial literature as positively linked to an active form of coping against adversity, even if unrealistic and ethical questions had been raised previously to learn about its relevance for cancer genetic risk practice [75].

### 2.4.5. Between duty, altruism, and intergenerational ethics

The orienting meanings and values assigned to genetic counselling embrace a sense of utility and protection (enabling disease prevention and health management), along with hope regarding the hereditary disease course of action. Altruistic values attached to the possibility of contributing for family medical care, especially for the sake of the younger generation, reveal an intergenerational ethics that shapes the engagement of families in genetic counselling, i.e., personal risks are linked to a web of cross-temporal, moral-relational connectedness to kin, also wrought by gendered-laden roles [76]. This anticipated and virtual caregiving for the sake of significant others enfolds a

distinctive moral landmark within the relational context of the interviewed families. Such ethics is at the core of individuals who are at (potential) risk, perceiving engagement in risk assessment and its management options as a responsibility. This context encloses a moral duty towards kin, since the at-risk individual acts as a *self in relation*, not only by sharing genetic information, but also by acting upon such information through engagement in some form of risk management [7, 76]. The same is applied to participants' involvement in prophylactic risk reducing interventions or/and surveillance screenings. These aspects, similarly to the role played by optimism and positive attitudes, showed the components of the whole risk context in which participants could actually do or decide something on their behalf concerning their possible futures.

### 2.4.6. Post-questionnaire data

From the post-questionnaire data one might infer that interviews have provided, to some extent, an opportunity for families to feel bonded and hear each other's accounts on a shared issue. The idea that the family interview allowed them to talk about difficult matters also emerged, as well as the opportunity to become more elucidated regarding the information available; this data may indicate that somehow the interview helped families to feel more enabled to manage their cancer susceptibility context. However, some participants reported intrusiveness, suggesting the interview's potential to activate difficult emotions. As families need to develop a sense of mastery in the face of adversity, such as cancer inherited risk, one can think that "giving information" for a study may function as an empowerment act for families [9].

### 2.4.7. Implications for practice

These findings may have direct implications for clinical practice. To our knowledge, cancer genetic counselling has incorporated family dimensions, although as adjunct services or sporadic supportive interventions. It is relevant to acknowledge and incorporate family-oriented data as part of ongoing psychosocial support throughout the genetic counselling protocol [3]. Interactions between individual and family development, individual and family autonomy, and kinship rights and obligations need to be considered for genetic counselling planning, including practice, and also for professionals training and policy making [6, 30, 41, 75]. The family context and history is relevant to participation in genetic counselling, testing, and surveillance methods. Therefore, it may also support families in the way they share information about genetic risk among relatives, providing professionals with tools that might allow them to assist consultands in their endeavour without jeopardising their professional liability. It is of great importance to consider other strategies to disseminate technical information among family members identified as potentially at-risk, for example through informative sheets, family letters, or CD-Roms. Furthermore, including a family

perspective may enable genetics healthcare practitioners to better understand how clients interpret genetic information, and the interactive patterns that inform common decision-making processes within cancer risk counselling. In addition, it will also help them learn about the subjective meanings and values that families assign to their risk. As many others have outlined [77-79], using narrative methods as a way to stimulate a family to jointly tell a more coherent story about its difficult experiences, such as illnesses, may have a rather transforming effect.

### 2.4.8. Study limitations

The fact that participant families were heterogeneous regarding their familial cancer susceptibility type (HBOC, HNPCC, and HDGC) makes it difficult to envisage the extent of accuracy of the implications of this study for each specific population. In addition, individuals also differed in their risk rates, arguably a factor which potentially draws specific features related to risk management within families, and how they perceive genetic counselling and their at-risk context. Furthermore, participant families were typically cohesive, meaning that the present study does not consider the experiences of less cohesive families, who arguably do not accept or manage to engage in a study addressing family interviews. It is likely that participants may have contacted those family members with whom they had a good relationship, whom they anticipate as being willing to participate due to lesser difficulties in discussing these themes. So, we must be contained in not generalising these results to all families involved in cancer genetic counselling.

This study relied on the index-patient's desire, availability, and capacity to inform other family members to participate in this family interview. Such process is similar to the cascade-screening method commonly employed in cancer genetic counselling, i.e., systematically approaching relatives of patients affected by genetic conditions and offering a predictive DNA test to those potentially at risk [80]. Cascade-screening is controversial and involves ethical objections, namely regarding autonomy and privacy issues. Debates about the right relatives have to know or not to know balance between the principles of non-maleficence, justice and reciprocity regarding the preventive value of the information and its potential for psychological harm [80, 81]. Although at a different level, it can be hypothesised that similar issues have been addressed by this investigation as well, namely because it also elicited moral dilemmas regarding whom (not) to tell, especially for those who chose to participate.

Research does not operate in a vacuum; it affects participants emotionally. As reported in the post-interview questionnaire, participants stated the emergence of intrusive or painful feelings during the family interview. It would have been important to provide families with access to a psychosocial health professional after the interview, in case they needed, so that any personal or

family issues unintentionally triggered during the interview could have been discussed with someone able to give appropriate support.

## 2.5. CONCLUSION

In conclusion, families' understandings of the experience of going through genetic counselling emerged from this study, namely, how risk awareness is elicited, its relation to action (motivators, risk management options) and consequences for the lives of families (psychosocial and developmental impacts), and the pathways by which cancer risk is tracked within families. In addition, families described how they attempt to integrate risk in their lives, and which values and meanings orient their experience. Families conceptualised cancer as a familial illness; positive genetic test results seemed to confirm family narratives embedded in transgenerational beliefs about the prevalence of cancer in their families, while altruistic values and a moral-relational duty seemed to inform about the familial engagement in genetic testing.

Familial experiences of genetic counselling and at-risk status encompass a sense of trajectory [19]. Familial experiences took the form of an historical and intergenerational narrative process, linking past, present and possible futures, and implying an ongoing set of individual and interactional experiences over time, rather than a consecutive order of stages. Such experiences are mediated by specific changes in the course of the illness (genetic test results, surveillance data, prophylactic interventions, symptoms onset, and hospitalisations) and by individual and family developmental lifespan transitions (marriage, childbearing, or death), conveying a complex interweaving of continuity and transformative paths for families in their process of risk accommodation, decision-making, and developmental tasks.

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## 2.6. APPENDIX

**Post-interview questionnaire** (adapted from Sobel & Cowan, 2000)

The family interview about the genetic counselling experience on your family was (check one):

___ a) emotionally neutral. I provided the information I expected I would.

___ b) helpful. It helped me to review and to clarify ideas and to identify some issues I had not thought of before.

___c) painful. It was uncomfortable for me to re-examine experiences connected with the genetic testing and the sharing of those results.

___d) intrusive. I was asked questions which were irrelevant and private.

Format: Being interviewed with the family members was (check one):

___a) a good idea. Explain.

_____

_____

___b) was not a good idea. I would have preferred that family members were interviewed separately. Explain.

_____

_____

Comments. Please include any comments that you feel would be helpful for the interviewer to consider in meeting with other families.

_____

_____

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# CAPÍTULO II

## INTERVENÇÃO PSICOEDUCATIVA: ABORDAGEM MULTIFAMILIAR

# 3. FAMILY MATTERS: EXAMINING A MULTIFAMILY GROUP INTERVENTION FOR WOMEN WITH *BRCA* MUTATIONS IN THE SCOPE OF GENETIC COUNSELLING[27]

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**ABSTRACT**

The availability of family-centred services for women genetically at-risk for breast and ovarian cancer (*BRCA*) due to deleterious genetic mutations is still scarce, despite the distress that these women and their families may experience. This study describes a multifamily group intervention for *BRCA* mutation carriers and their families. Methods include a time-limited psychoeducational programme involving educational and support components, and consisting of four semi-structured multifamily sessions. Three families (a total of 9 people) attended the programme in genetic counselling for hereditary cancers at a Portuguese public hospital. A focus group interview was performed to assess both the practical and the psychosocial impacts, and to collect suggestions from participants. The present paper focuses on the practical aspects of the intervention, its development and its evaluation. Participants reported that the programme is well-structured and that responds to the needs of patients and their families by improving coping skills and medical awareness in the adaptation to genetic illness. Results reinforce the need to integrate psychosocial and family-oriented interventions in genetic counselling, addressing the holistic experience of hereditary disease. Recommendations for enhancing the services available are provided. The multifamily discussion group, combining educative and supportive services with a family focus, can be successfully adapted in genetic counselling protocols.

**Keywords:** genetic counselling, hereditary cancer, psychosocial genetics, psychoeducation, multifamily groups.

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## 3.1. INTRODUCTION

As genomics continues to expand its expertise in cancer genetics, scientists and clinicians are given the chance to learn more about the illness mechanisms and the available preventive or prophylactic treatments. For those who face an increased genetic risk due to inherited deleterious gene mutations, it is crucial to be able to accommodate this rapidly changing knowledge in their health management and into their lives (Rolland and Williams 2006).

Genetic counselling examines family medical histories to educate clients about their risks for an inherited disorder and the options for dealing with the risk of occurrence (National Society for Genetic Counselors 2006). Cancer risk counselling has grown rapidly in recent years to become a major area of specialisation within genetic counselling. Its main purpose is to evaluate the tumour risk in families whose members have a hereditary predisposition for cancer and to inform them about treatment options and preventive strategies enhancing their ability to make informed decisions (Sifri, Gangadharappa and Acheson 2004). These are the typical guidelines for genetic counselling in hereditary tumours, according to the guidelines adopted at the genetic centre where our study was developed: a) collection of family and individual medical history; b) genetic risk assessment; c) predictive testing for genetic susceptibility to cancer; and, d) surveillance and preventive screening, treatment and follow-up sessions.

In Portugal, genetic services are generally integrated in the National Health System (NHS), mainly in central hospitals, but also public institutes (partially integrated in the NHS), some medicine faculties, and private entities (often assisting with clinical and laboratorial expertise). Cancer genetics services are available in regional oncological hospitals, and included under the umbrella of central hospital departments. A National Program of Pre-Symptomatic Test and Genetic Counselling was approved and implemented in the mid nineties, primarily directed for adult neurological onset disorders, and later expanded to other forms of genetic disease (Health General Directory 2004).

Genetic counselling for hereditary tumours commonly exposes the client to a significant degree of uncertainty and various levels of psychological distress, encompassing diverse practical and emotional challenges (Evers-Kiebooms et al. 2000; Pasacreta 2003). The psychological effects of being at increased risk for breast and/or ovarian cancer have been extensively studied, pointing to a frequent experience of distress (Schlich-Bakker, Ten Kroode and Ausems 2006). A vast body of literature illustrate the social and emotional effects of genetic conditions, with potentially negative impacts in individuals and families (McAllister et al. 2007; Patenaude 2005). Distress among *BRCA*1/2 carriers fluctuates over a 5-year period, and depends on the degree of integration of genetic information attained by individuals (Werner-Lin 2008).

Genetic counselling is increasingly recognized as not solely an individual, but also a family matter that poses a complex interweaving of biological and interpersonal interactions across nuclear and extended family members (McDaniel 2005; Peterson 2005). Families exposed to increased genetic risk tend to define their identity based almost exclusively on that experience; this reorganization around the ambiguous territory of at-risk status, and the need to develop coping efforts under emotionally charged contexts, is suitable to disrupt key aspects of family functioning, giving the family an identity as a *genetically ill family* (Patterson and Garwick 1994; Sobel and Cowan 2000).

Despite the integration of psychosocial models has been stated as an essential feature of patient care in genetic counselling services (Cappelli et al. 2009), as well as the need to produce investigation reflecting the use of psychosocial support by families (Douma et al. 2008), standard care in such settings usually comprises a predominantly biomedical oriented protocol, without ongoing psychosocial or family-based support, especially after genetic testing results are delivered (Werner-Lin 2008). Therefore, it becomes crucial to develop family-oriented models of coping, adaptation, and integration in inherited risk management, such as multifamily discussion groups (McDaniel et al. 2006).

This paper describes a psychoeducational programme for women carrying a *BRCA*1 and/or *BRCA*2 mutation, and their families, in the scope of genetic counselling during the post-test period. The programme design and the session's contents will be reported, as well as its evaluation through the participants' views on the structural and practical aspects of the programme, and the perceived benefits in their lives. In addition, recommendations will be provided on how to adapt further family-oriented psychoeducational interventions in the scope of genetic counselling for hereditary cancers.

### 3.1.1. Conceptualising psychoeducational interventions in cancer genetic risk

Recent decades have witnessed the significant development of psychoeducational models, which integrate health education and psychosocial support into a multidisciplinary perspective, aiming to: increase the family and patient's sense of competence in dealing with the illness; promote working partnerships between health professionals, families and patients (McDaniel et al. 2005).

Psychoeducational approaches are shown to be more efficient if developed in multifamily discussion groups (Ostroff et al. 2004); this context promotes a non pathological atmosphere, allowing families to learn and talk about the illness, namely about the way it *invaded* the family identity and how it interferes in its development. Its effectiveness is recognised, in particular because (Ostroff et al. 2004): it reduces family stress; it enhances the sense of competence amongst patients and their families; and it increases their adherence to treatments. Multifamily discussion groups are time-limited (4-6 sessions); they are especially designed to help family adjustment and

coping with illness demands and uncertainty, allowing support from people who are sharing the experience and relieving feelings of isolation (Gonzalez and Steinglass 2002). Such interventions have the potential to be integrated into the genetic counselling protocol, since they address the psychosocial interface between medical, individual and familial issues.

Psychoeducational interventions may provide support for individuals and families at-risk for hereditary cancers, by improving mastery and resilience at key points in the illness adaptation process (Werner-Lin 2008). Karp et al. (1999) reported a 6-session support group for *BRCA* carriers, addressing the issue of prophylactic mastectomy, and highlighting intervention benefits and cost-effectiveness as a valuable adjunct procedure to individual genetic counselling. Another six-week psychoeducational group for women at high genetic risk for *BRCA* was designed by Speice et al. (2002), addressing core themes and providing recommendations to meet the needs of affected families. A six-month supportive-expressive group intervention was used to address the medical and emotional needs of *BRCA* mutation carriers (Esplen et al. 2004). Such programmes, however, do not include other family members (besides the patient), who may also benefit from participation in these programmes.

## 3.2. PLANNING

### 3.2.1. Programme design

This programme was designed based on three main sources: i) existing literature and research on genetic counselling for *BRCA* mutation carriers, and psychoeducational interventions in medical settings; ii) a previous study developed by our team on the experiences of clients undergoing genetic counselling for hereditary cancers, in which participants reported that there should be made available to them interventions focussing on the following aspects: improving clients' psychosocial adjustment; providing medical education and counselling, given their need to understand genetic information; supporting individuals and their families in the decision making process, and assisting them in coping with the demands of genetic illness (Mendes, Santos and Sousa 2010); iii) needs reported by the women at-risk for *BRCA*; during the recruitment, potential participants were interviewed about their psychosocial needs and the topics they would like to see included in the programme; participants mentioned the following: medical information about *BRCA* mutation and its implications; preventive and prophylactic treatments available; coping strategies for promoting well-being; and psychological support both at a personal and familial level.

### 3.2.2. Participant's recruitment

Recruitment is a crucial stage, in particular because families that show enthusiasm at the first contact may afterwards give up (Ransom, Azzarello and McMillan 2006); the abandonment rate in

this study was 25% (one family); this family, in comparison to the participant families, comprises a larger number of members (especially siblings) and a more accentuated geographical dispersion of their residencies.

Following approval by the Ethics Committee, participants were selected at the Hereditary Tumours Consultation of the Centre of Medical Genetics and Human Reproduction of the University Hospital of Coimbra (Coimbra, Portugal). Recruitment criteria were based on the following: at least one member of the family was supposed to have tested positive for deleterious BRCA1 and/or BRCA2 mutation. In this study all family members were eligible to participate in the group, whether or not they had done genetic testing, and whether or not they were mutation carriers, because genetic risk is a family issue that emotionally affects all family members, men and women, either related or not by bonds of consanguinity (McDaniel 2005; Peterson 2005).

The recruitment process involved the following steps: searching through the service database for eligible participants; making a phone call to present the aims of the study to participants and ask for their collaboration with assured confidentiality; sending letters to those users who agreed to participate, in order to confirm the previously given information and make the same information accessible to other family members, and to provide them with an informed consent form to be delivered at the first session; a second phone call was made to deal with practical issues concerning the scheduling of sessions (all were scheduled ahead of time), including a previous family interview; finally, family interviews with the potential participants were made (mostly at the participants home or at the hospital) to collect demographical and psychosocial data, and to assess needs.

### 3.2.3. Participants

One group was set up involving 3 families and 9 people. Participant families included 2 to 4 elements (Table 3.1.). Participants were all females linked by blood kinship, including sisters, daughters, grandmothers and aunts (all participants had carried out the genetic test and had increased risk for breast and/or ovarian cancer). The only exception was the husband of a participant, who attended the last session.

**Table 3.1. Participants' demographics and cancer history**

| **Participants' demographics, illness features and family history** | ***n* = 9** |
|---|---|
| Mean age | 43.5 (24 – 74) |
| Sex | 9 females |
| Race | 9 Caucasians |
| Marital status | |
| Married | 7 |
| Single (Dating/In a relationship) | 1 (1) |

| | |
|---|---|
| Mean years of education | 11 (4 - 20) |
| Mean number of months since last genetic testing | 21.6 (8 – 36) |
| Genetic risk status | 9 *BRCA* mutation carriers |
| Personal history of cancer | 4 affected (1 symptomatic) |
| Mean number of cancers in the family | 5.66 (2 -8) |
| Mean number of at-risk individuals in the family | 4.33 (2-7) |
| Mean cancer-related deaths in the family | 2 (1 - 3) |
| Number of participant families | 3 |

**Table 3.1. Participants' demographics and cancer history** (cont.)

### 3.2.4. Procedure

The programme was implemented in a multifamily discussion group format. There were four semi-structured sessions (Table 3.2.), coordinated by two facilitators, with training in medical family therapy and with previous experience facilitating multifamily groups for patients facing cancer (Chiquelho et al. 2010). Facilitators adopted an active and empathetic posture, in order to assist participants in normalising feelings, reinforce family competencies and resources, and mediate the interaction between group participants.

Each session was expected to last 90 minutes, but time invariably extended to 120 minutes. All sessions were videotaped with the permission of all participants. The sessions were scheduled at the participants' convenience at weekends (Saturdays, 10:30am-12:00pm), and they were free of charge for participants. The programme comprised two components: i) education, aiming at providing up-to-date medical information about BRCA mutations, prophylactic treatment options and community based resources; and ii) support, including disclosure and family reactions to testing, emotional reactions and coping strategies, and family identity maintenance. Each session had the flexibility needed to integrate themes that spontaneously emerged from the participants.

**Table 3.2. Programme summary**

| Session | Format/Segment | Contents |
|---|---|---|
| 1 | Presentations | Presentations. |
| | Support | Impact of increased genetic risk in the family. |
| 2 | Education | Medical information. |
| | | Community based resources. |
| 3 | Support | Embracing family identity |
| 4 | Support | Problem-solving techniques. |
| | Ending | Stress management strategies. |

#### *3.2.4.1. First session*

The aim of this session was to explore the impact of genetic risk awareness in the family. Initially, the need to maintain confidentiality among participants was underlined, and permission to videotape obtained. After presentations of both participants and facilitators (who also introduced the program format), participants were gathered by family and encouraged to share the negative and positive aspects that had emerged. It was emphasised that such knowledge is commonly perceived as alarming; however, thinking about family functioning opens the possibility of recognising the significant aspects of such an experience. The positive aspects highlighted by participants were family union and affective strength, whilst the negative aspects included the face of uncertainty in the future, excessive concern with symptoms, and increased anxiety and distress. Next, families were invited to think about how they could overcome the negative aspects, having mentioned that such effort may be achieved if closeness and solidarity among family members is improved, and if access to medical education is provided. Conclusions were shared among all participants. This was an emotionally intense phase, creating thus empathy among participants and facilitators. Finally, a home task was requested: each family was asked to gather and discuss their doubts and concerns about their genetic risk condition, in order to discuss these with the doctor, who should be present at the following session.

#### *3.2.4.2. Second session*

This session aims to provide information about the medical aspects of *BRCA*, to dissipate doubts, and allow for a better genetic illness management. A doctor (geneticist) was present in the session in order to provide up-to-date medical information on *BRCA*1/2 testing, treatment options and related concerns, as well as to answer participants' questions and talk about their doubts. The following themes were addressed: genetic and hereditary illness significance, mechanisms of hereditary transmission of genetic susceptibility, and prophylactic treatment options for genetic risk management; some community based resources were also made available, such as Internet networks and forums. Then, participants discussed their doubts with the doctor; the more recurring query topics were: medical surveillance procedures, reproductive and childbearing issues, latest technological advances in genetics, informative resources and preventive measures for descendants. Participants were actively involved in this session; they considered it was easier for them to ask questions to the physician in this setting.

#### *3.2.4.3. Third session*

The main goal of this session was to help families keep a sense of continuity in their own history and identity (past – impact of genetic risk, present – current challenges, and future – upcoming

projects). The session started with a brief explanation about how a crisis like the threat of genetic risk for cancer may affect families, creating a gap between the family's past identity and the frightening present, and putting the future on hold. Aiming to explore how genetic risk shaped the sense of family identity, facilitators invited each family to think about their values before, during, and after the genetic positive test result, asking them to disclose about the family functioning main features in each period. Families stated that their values remained the same across time, although the involvement in genetic counselling and the *arrival* of testing results had increased solidarity and unity among family members, including extended family and significant others. In this session participants reported a sense of vagueness, as well as problems with understanding clearly what they were meant to do or think about; facilitators shared the same opinions as participants regarding this matter.

#### *3.2.4.4. Fourth session*

The last session aimed at helping the participants develop stress-management strategies, since high levels of stress and anxiety are commonly reported amongst genetic illness patients. Two types of coping strategies were addressed: problem solving and relaxation techniques. Concerning the first strategy, facilitators stated the universal nature of experiencing problems when facing the challenges of an illness, or of a potential illness, and then asked each family to share some of their current problems; a problem solving exercise was developed based on decisions concerning involvement in prophylactic surgery, following these steps: identifying the problem; naming it; sharing points of view; finding possible solutions; deciding on one alternative; carrying out that decision; monitoring to evaluate its effectiveness (McDaniel et al. 2006). As for the stress management, the normative character of stress was underlined through examples that showed how stress becomes "natural" in certain circumstances of uncertainty. Cognitive and progressive muscle relaxation training were introduced and performed as an additional resource to the strategies they could use to manage stress.

### 3.2.5. Programme evaluation

#### *3.2.5.1. Objectives and methodology*

The programme was evaluated through a focus group semi-structured interview (Krueger and Casey 2000), in order to: collect opinions and suggestions from the participants to better adjust the programme to their needs; identify the individual and family benefits/impacts. The purpose was to improve the programme and make it suitable for incorporating the genetic counselling protocol for hereditary cancers as a complementary family-focused tool to address a better psychosocial adjustment. This qualitative method was chosen because (Krueger and Casey 2000): it is pertinent

for exploratory approaches, since it is a rich method of revealing experiences and perceptions; it is appropriate for sensitive topics, which is the case of an intervention addressing genetic illness experiences, as it provides a safe environment for participants to share their thoughts and feelings.
The focus group took place one month after the last session, and was conducted by the programme facilitators. The interview was approximately 90 minutes in length, and was videotaped, transcribed and submitted to content analysis. All participants agreed to cooperate. The interview was applied according to the guidelines for conducting focus groups (Piercy and Hertlein 2005), comprising the following topics: the functional aspects, contents and activities of the programme; individual and family benefits; suggestions to increase the sense of well-being.

### 3.3. RESULTS

All participants considered that all functional aspects of the programme were adequate (number of sessions, duration and frequency), as that they prevented dispersion without being a burden to personal and family life:

> *"I think the program was well planned; the once-a-week format was ideal, otherwise some people would probably have to leave other things behind"*. [Rita[28], 28 years old]

The way sessions were structured and its contents were pointed as useful because it focused on specific topics and encouraged the conversational engagement of all participants. Nevertheless, some deviation from the given proposed subject has occasionally emerged (3/9):

> *"I liked the balance between working on a specific topic and sharing our stories freely without rigidly limit our conversation"*. [Andrea, 25 years old]
>
> *"Sometimes I felt that the group had dispersed from the topic we were discussing; but I agree that such 'freedom' was very important for us all, otherwise we probably had stayed more passive"*. [Rita, 28 years old]

The multifamily discussion format was highly approved and one of the key benefits for participants', since it allowed the creation of an atmosphere which encouraged the sharing of personal and family experiences:

> "*Sharing this problem with other people facing the same is very important; of course I can talk with friends, but here we were all connected with this issue"*. [Isabel, 23 years old]

The main benefits were centred on the group experience of sharing, which removed the inadequacy of some feelings and thoughts and prevented isolation. A sense of closeness also appeared as significant:

[28] All participants´ names were changed for confidentiality purposes.

> *"The opportunity to exchange experiences and listen to stories from other people facing the same difficulties made me feel that I was not alone in the world, with my worries and fears... I'd never met anyone that had the same problem!"*. [Ana, 36 years old]
>
> *"For me it wasn't just a matter of sharing; here we could feel close and bonded to each other… despite our differences we were very similar, in our needs, fears, doubts…"*. [Maria, 53 years old]

The access to simple coping strategies enabled a positive integration of the at-risk status in participant's lives (2/9); differences were acknowledged in the way they were *thinking* and *seeing themselves* about living with increased genetic risk:

> "*I feel that now I have strategies to live better with this* [increased genetic risk]; *I am able to face it not as a deficiency, or incapacitation, but as something that is part of me and that I carry with myself everyday...*". [Andrea, 25 years old]

All participants felt the informative session as very important, allowing an effective integration of medical information because it was easier for them to ask questions to the physician in this setting:

> "*The second session was central for me; questioning the doctor directly about our doubts was very important because during the medical consultations we used to be very anxious and couldn't process everything…"*. [Sofia, 47 years old]
>
> *"This field* [genetics] *is constantly evolving and changing; that session updated what I knew since when I undergone testing, four years ago"*. [Ana, 36 years old]

An increased self-assurance when considering decision making about undergoing prophylactic surgery treatments was also mentioned as a benefit (4/9); two women reported an improved confidence to undergo risk reduction procedures such as oophorectomy and subcutaneous mastectomy:

> "*I used to postpone because I was really scared to face all the necessary medical steps for prevention (which I will, now I can say it), and now I feel more secure and confident about it*". [Andrea, 25 years old]

The oldest woman in the group shared her relief to see that the family accepted to participate in these sessions:

> "*This family has already suffered great pain with losses, and I fear for my daughters and for my granddaughter; I am glad because I feel that they are more relaxed and confident"*. [Claudia, 72 years old]

Participants' reported the facilitator's role as adequate, namely their interest in "*equally listening to us all*", and the use of practical strategies "*focusing not only in our personal experience, but also in family relations*". Yet, a more directive approach was mentioned as potentially adequate in order to avoid thematic dispersion (2/7):

> *"In some occasions perhaps you* [facilitators] *could have been more directive, preventing some 'side-talking'…maybe by establishing a more formal structure, like limiting our time to talk…"*. [Rita, 28 years old]

When asked about the negative aspects of the programme, two participants pointed some parts of the third session; albeit considering it "useful", they mentioned a sense of vagueness regarding what they were meant to do or think about; facilitators shared the same opinions as participants on this matter:

> "*I got the sensation that in the third session we ended up talking about other things that weren't what you had planned…"*. [Ana, 36 years old]
>
> *"I felt that we had done something very similar to what we'd already done in a previous session* [first session]*…*". [Isabel, 23 years old]

As for suggestions to improve the programme, one participant recommended the inclusion of a "*physical liberating and cathartic ritual*", allowing participants to actively express feelings of anger and despair, mainly because others (2/9) felt that relaxation techniques were not suitable for them. Two of the younger participants suggested, as a complement to group sessions, the availability of an individual therapeutic setting centred on more personal issues. Participants (6/9) also suggested the inclusion of other professionals to provide medical information within the domains of plastic surgery, radiotherapy and gynaecology/obstetrics. For this purpose, an additional informative session might be considered:

> *"A surgeon, and maybe a radiologist, could also be present; it would be useful for those who have doubts regarding mammary reconstruction..."*. [Sandra, 45 years old]

### 3.4. DISCUSSION

This qualitative exploratory study provide insight on the adequacy of the intervention previously described in helping patients and their families to adjust to the challenges of increased genetic risk for BRCA. Results suggest that the programme it is well-structured, taking into account its duration, contents and the methodologies used, and that it meets family needs in terms of adjustment to face a genetically linked condition.

Regarding the practical aspects of the programme, it would be useful to consider the redesign of the third session. As both participants and facilitators reported a sense of vagueness and similarity with a previous session, although acknowledging its relevance, we suggest addressing the families' identity through another configuration. Therefore, by asking families to think about objects suitable of symbolizing families' values across time (such as photographs, or material items inherited through generations), and to bring them to the session in order to evoke and share its meaning, families are thus encouraged to recall stories (Trees, Koenig Kellas and Roche, 2010) based on

central aspects of family life that may have been shadowed by the current distress associated to genetic illness issues; this way it seems possible to activate the creation of a narrative promoting a sense of continuity in their own history, and embracing families' identity.

As for the psychosocial impact of the programme, the multifamily group format represent an opportunity for participants to listen to others experiencing similar circumstances, enabling emotional expressiveness; the group format helped to enact a mutual, supportive atmosphere that allowed the normalisation of feelings about genetic risk. Also, experiencing bonding and extra-familial networks seemed to have contributed to relieve the psychosocial burden at both personal and family levels. In addition, this programme helped to create a sense of control over one's health management, as the session with the clinician provided a chance to increase medical awareness regarding genetic information, and consequent self-assurance on decision making about prophylactic and risk reduction treatments.

Through the reciprocal process of listening and narrating their medical family history, as well as their identity values, families were allowed to revise and frame stories about genetic risk, helping to create a sense of mastery and empowerment concerning living with a genetic condition. Overall, this programme seems to provide a useful tool towards the incorporation of family-centred interventions in follow-up support for BRCA at-risk individuals and their families, complementary to the more biomedical oriented and person-centred protocol of genetic counselling.

### 3.4.1. Recommendations

The following suggestions may enhance a family-centred focus on genetic counselling; these topics are suitable to address individual and family needs, as more research data becomes available:

i) Integration of a professional trained on psychosocial genetics in the genetic health care team to assess routine concerns and provide support to clients and families;

ii) The availability of psychoeducational multifamily support groups, throughout genetic counselling protocols (pre- and post-testing);

iii) Incorporation of participants' suggestions in the multifamily groups structure, such as the inclusion of a plastic surgeon in the informative component, the integration of diverse relaxation techniques, and the access to an optional individual therapeutic setting as a complement to group sessions for people who do not feel comfortable in group or family settings.

iv) Referrals to mental health professionals in cases of persistent psychosocial distress or related symptoms.

These multidisciplinary family-oriented recommendations call for collaborative work among different health care providers, in order to develop innovative psychosocial interventions

addressing the holistic needs of those seeking help from genetic counselling services (McDaniel et al. 2005; Speice et al. 2002; Werner-Lin 2008).

### 3.4.2. Limitations and research perspectives

Further research should include a larger sample and be carried out in other genetics services to evaluate the applicability of the presented programme. It would also be useful to perform focus-groups amongst genetic health-care practitioners examining the strengths and limitations of incorporating this intervention in the mainstay of genetic services, and what health professionals would be involved. Research should also produce data regarding the adaptability of this intervention, namely in the pre-test phase and immediately after disclosure of genetic test results, adapting the programme contents to the psychosocial characteristics of each phase (Rolland and Williams 2006).

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# 4. EMBRACING FAMILY SUPPORT IN GENETIC COUNSELLING: A MULTIFAMILY DISCUSSION GROUP FOR COLORECTAL CANCER SUSCEPTIBILITY FAMILIES[29]

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## ABSTRACT

A non-experimental exploratory study examining a multifamily discussion group for colorectal at-risk individuals and their families, in the scope of genetic counselling, is described. Four families attended a 90 minutes four-session programme at a genetics centre of a Portuguese public hospital. A post-programme focus group interview was performed to assess practical and psychosocial impact, and collect participant's views. Participants reported that the programme responds to the patients and families needs, enhancing their adaptation and coping to genetic illness. Results reinforce the need to integrate family-centred interventions in genetic counselling services, addressing the holistic experience of hereditary disease. We conclude that the programme provides an integrated health care setting to help at risk individuals and their families cope with the specific biopsychosocial demands of their increased genetic susceptibility.

**Keywords**: chronic physical illness, medical family therapy, multiple family approaches, psychosocial & psychoeducational approaches, genetic counselling.

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## 4.1. INTRODUCTION

Recent innovations in the field of genetics and genomics opened diverse possibilities into the diagnosis, treatment and prognosis of several illnesses, posing a range of challenges for practitioners and individuals involved in genetics health care. Hereditary genetic testing can, for example, detect familial conditions and risks, with implications that rarely affect just one individual in the family (Featherstone *et al*., 2008; Gaaf and Bylund, 2010)

Biological relatives share genetic material that is suitable to determine of of developing a particular condition themselves or of transmitting such inherited genetic mutations to offspring. Consequently, the presence of an inherited condition affects not only the biological family members who may or may not be at risk, translating social relations and categories into biological terms (Atkinson and Glasner, 2007). These "risky relations" have implications for blood kinship, but also for couples, particularly for their reproductive planning, and for the extended family as a support system, who may be involved in the myriad of complex issues posed by increased genetic risk (Featherstone *et al*., 2008).

As genetic risks are distributed among family networks, becoming aware of an increased genetic susceptibility to cancer can be a disturbing event, both for personal and family levels (Evers-Kiebooms *et al.*, 2000). Genetic counselling and testing research focusing on families has grown during the past decade (McDaniel, 2005; McDaniel and Campbell, 1999; Rolland, 1999; Sorenson and Botkin, 2003), after psychosocial genetics had emerged with a wide range of themes outside the exclusive biomedical approach (Harper, 1993). All these developments have contributed to the need to examine genetic predisposition and the genetic counselling experience from a family or broader social context perspective (Peterson, 2005; Van Riper, 2004); genetics is recognized as not solely an individual and biomedical matter, but also as a familial and psychosocial one, and its practice as not just with and about individuals.

A family view on genetic counselling highlights the need to understand the psychosocial demands for those who face an increased genetic risk, as well as to develop adequate family-focused interventions in the scope of genetic counselling (Cameron and Muller, 2009; Wang *et al.*, 2004). Although research shows the distressing potential of genetic counselling and high genetic susceptibility (Evers-Kiebooms *et al.*, 2000; McAllister *et al.*, 2007; Meiser, 2005), and despite stated as a crucial feature of patient and family care in genetic counselling services (Cappelli, *et al.*, 2009; Douma *et al.*, 2008) the availability of psychosocial support for at-risk individuals and their families in such context is still scarce (Cappelli *et al.*, 2009). This article describes a multifamily intervention programme for colorectal cancers (CRC's) predisposing mutation carriers (hereditary non-polyposis colon cancer, HNPCC, and familial adenomatous polyposis, FAP), and their families, in the scope of genetic counselling, during the post-test period. The programme design

and session contents are reported, as well as its evaluation through the participant's views on the programme's structural and practical aspects, and the perceived benefits on their lives.

### 4.1.1. Profile of CRC's gene alterations

CRC's are serious life-threatening conditions requiring adequate and life long clinical surveillance. The invasive treatments that are often needed, and its early age of onset, create a demanding context for those facing CRC's gene alterations and for their families. While most of CRC's are "sporadic" (65-85%), 10 to 20% of these types of cancer are familial or hereditary, meaning that inherited mutations lie as the primary risk factor (Helm *et al.,* 2003). The most common forms of CRC are hereditary non-polyposis colon cancer (HNPCC) and familial adenomatous polyposis (FAP); HNPCC accounts for at least 2% to 3% of CRC's (Allen and Terdiman, 2003), while FAP accounts for 1% (Meropol *et al.*, 2006). In each of these syndromes, multiple primary tumours and early-age onset are characteristic features. Individuals with a HNPCC mutation have an estimated 30-74% risk of disease manifestation before the age of 70, and an additional increased risk of other HNPCC-associated tumours (endometrium, stomach, biliary tree, ovaries and renal pelvis). People at risk are advised to undergo biannual colonoscopy between the ages of 20 and 25, and should consider endometrial screening beginning at age of 30 to 35 years (Helm *et al.*, 2003). FAP is characterised by the early development of multiple adenomatous polyps in the colorectum from the age of 10 onwards; without preventive measures, cancer risk before the age of 40 is almost 100%% (Meropol *et al.*, 2006). Prophylactic surgery is usually offered between the ages of 15 and 25 years. Prophylactic measures for those diagnosed with or at risk for FAP include regular endoscopic surveillance at age 12-35 years; if a high number of polyps are found, prophylactic colectomy is indicated total of 50-90% of patients will also develop duodenal adenomas, and 10-15% of the patients will develop desmoid tumours, with increased risk of developing additional life-threatening problems (Scott, 2003).

### 4.1.2. Genetic counselling for hereditary cancers

Cancer risk counselling evaluates the tumour risk in families whose members have a hereditary predisposition for cancer, and informs them about treatment and preventive strategies enhancing their ability to make informed decisions regarding their health management (Sifri *et al*., 2004). General procedures in the scope of cancer risk counselling generally include (Trepanier *et al.*, 2004): a) collection of family and individual medical history, using family pedigree information to assess genetic susceptibility, as well as other relevant personal and familial medical information (such as cancer diagnosis, age of onset, and anatomic-pathological data); b) genetic risk psychosocial assessment, aiming to appropriately inform clients about their genetic risk and the

available medical surveillance procedures, and to assess current and potential psychosocial difficulties; c) susceptibility genetic testing for cancer, after genetic counselling and education, and under informed consent, aiming to search for a pathogenic mutation; and d) surveillance and preventive screening, treatment and follow-up sessions.

The outcomes of genetic testing for cancer-predisposing gene mutations may reveal the following (Feetham and Thomson, 2006): i) if the client carries a known gene mutation, already identified in another family member (positive result), there is an increased susceptibility to cancer, and testing can be offered to family members to determine whether they also carry the mutation; or ii) if a pathogenic mutation is not identified at mutation search testing (negative or indeterminate result), this may be because: the individual tested does not carry the index pathogenic mutation; he/she carries a variant of uncertain significance; a mutation was identified in another gene which has not yet been identified; or the individual tested do not have a genetic predisposition to cancer. Failure to find a mutation in an index case does not remove the possibility of cancer nor does it decrease other associated cancer risks.

### 4.1.3. A family-oriented lens on genetic illnesses

Genetically linked conditions are an intense biopsychological experience and a demanding crisis for a family system. The familial implications of genetic risk may be examined within the family system theory approach (Peterson, 2005), where events suitable to affect an individual also affect the whole family system.

A genetic diagnosis and its implications, as for the case of hereditary tumours, are viewed not as a single event, but rather as an ongoing, dynamic process that may have repercussions over time (Galvin and Young, 2010). Genetic conditions also pose a burden on relatives, whether the results are positive (mutation carrier) or negative (non-carrier); even those who are unaffected and not at risk may experience higher levels of anxiety, and survival guilt, after receiving a non-carrier result (Biesecker and Marteau, 1999). Families dealing with genetic illnesses are typically confronted with several practical and emotional challenges, often carrying significant uncertainty (concerning disease inheritance, timing of onset and symptoms severity, preventive and prophylactic procedures and timings, personal and family planning) and psychological distress (since procedures are permeated by anxiety, worry and doubt, and highly complex nature of genetic information (Evers-Kiebooms *et al.*, 2000; Mendes *et al.*, 2011). Families' with increased risk status may experiences disruption in key aspects of family functioning potentially becoming rigidified in their identity as a *genetically ill family* (Patterson and Garwick, 1994; Sobel and Cowan, 2000). Familial cancers commonly reveal multigenerational patterns of illness manifestation, and can shape families' processes, norms and expectations about the individual and the family life cycle. The interactions

between transitions occurred in individual and in the family' life cycle and the genetic counselling timeline assume particular clinical significance (Street *et al*., 2000).

A family resilience approach (Walsh, 1996) enables families to manage such demands by strengthening relational ties and coping styles, not only through problem solving or decision making, but also through problem prevention and preparing family members to meet future challenges. This systemic focuses also involves support networks and larger systems to promote community connections that families may have lost. Resilience is also promoted by contextualising and normalising the crisis, and by offering pragmatic guidelines for adaptation. It is generally accepted that family-oriented psychosocial interventions on genetic health care are a key tenet to address the immediate and long-term needs for those genetically at risk (McDaniel *et al.*, 2006).

Street and Soldan (1998), following Rolland's previous work on chronic illnesses (1994), envisioned the need to acknowledge the *pre-illness phase* in the illness time course. This is a relevant issue since in many cases mutation carriers live considerable time pre-symptomatically, before visible symptoms of the disease become noticeable. A particularly useful model when considering a family-centred approach of genetics is Rolland and Williams Family System Genetic Illness Model (Rolland and Williams, 2005). Deeply following a biopsychological approach to health (Bourne, 2010; Engel, 1977), and updating the original Family Systems Illness Model (Rolland, 1994), this model incorporates the growing knowledge of genetics in disease risk, manifestation, prediction and diagnosis, offering a framework to consider the psychosocial demands of several types of genetic illnesses. The authors emphasise the interaction of both symptomatic and non-symptomatic phases of a genetic illness, highlighting the nature of genetic conditions and the uncertainty often present (regarding the likelihood of developing the disease, timing of clinical onset, and treatment options) by describing a psychosocial typology in a developmental fashion. The updated model includes the experience of genetic disorders and genetic counselling in a timeline comprising four phases: i) awareness of possible genetic risk, involves seeking basic information about the illness and establishing initial communication in the family; ii) crisis I (pre-testing phase), encompasses the psychosocial understanding of the illness, the active consideration and decision regarding testing, and the information of family members; iii) crisis II (test/post-testing phase), involves the incorporation of the testing outcome into personal and family life, and the consideration of the prophylactic treatment options available; iv) long-term adaptation (if results are positive), refers to the balance between proactive personal and family planning and the need for up-to-date genetic information. The model also emphasises variable factors such as the likelihood of developing the condition, timing of clinical onset, and available treatment options, providing an integrative framework that furnishes the possibility of incorporating a family-oriented lens on genetic counselling.

### 4.1.4. A practical intervention tool: psychoeducational multifamily discussion groups

Recent decades have witnessed an increasing development of psychoeducational models (McDaniel *et al.*, 2005; Steinglass, 1998), which integrate health education and psychosocial support in a multidisciplinary perspective, aiming to: increase the families and patients sense of competence in dealing with the illness; and promote working partnerships between health professionals, families and patients.

Albeit the intense debate considering its effectiveness and potential moderators, psychoeducation appears as one of the key psychosocial interventions for cancer patients (Zimmermann, 2007). These approaches have shown to be more efficient if developed in multifamily discussion groups (Ostroff *et al.*, 2004), because it facilitates a context that promotes a non pathological atmosphere, allowing families to learn and talk about the illness, namely about the way it *invaded* the family identity and how it interferes in its development. Its effectiveness is recognised, in particular because (Ostroff *et al.*, 2004; Steinglass, 1998): reduces family stress; enhances the sense of competence amongst patients and their families; and increases their adherence to treatments. Multifamily discussion groups (Gonzalez and Steinglass, 2002) are brief and time-limited (4-6 sessions), psychoeducationally oriented, and mainly designed to help family adjustment and coping with illness demands and uncertainty, allowing support from people who are sharing the experience and relieving feelings of isolation (McDaniel *et al.*, 2006). Such interventions may improve mastery and resilience at key points in the illness adaptation, and have the potential to be integrated into genetic counselling protocols, since they address the psychosocial interface between medical, individual and familial issues (Rolland and Williams, 2005). Psychoeducational programmes for at-risk mutation carriers have been performed as a psychosocial tool concerning the prophylactic mastectomy dilemma (Karp *et al.*, 1999), a family-systemic orientation in genetic counselling (Speice *et al.*, 2002), or addressing the medical and emotional needs of breast and ovarian cancer (BRCA) mutation carriers (Esplen *et al.*, 2004; Mendes *et al.*, 2010). Such programmes, however, do not include other family members (besides the patient), who may also benefit from participation in these programmes.

## 4.2. METHODS

To address the issue of updating and supporting CRC susceptibility families, a multifamily discussion group was designed, implemented, and evaluated. A non-experimental exploratory study was carried out using qualitative methodology processes, including: individual and family semi-structured interviews and a focus group interview (Piercy and Hertlein, 2005), a grounded theory approach (Glaser and Strauss, 1967), and a nonrandomized intervention comprising the principles

of psychoeducation (Brown, 2004; Roberts *et al.*, 2002) and multifamily discussion groups (Steinglass, 1998).

### 4.2.1. Design

This programme is adapted from a range of multifamily interventions with a psychoeducational focus developed in cancer and other chronic illnesses (Chiquelho *et al.*, 2011; Steinglass, 1998), and based on the existing literature and research on cancer genetic counselling and its psychosocial implications (McDaniel *et al*., 2006). A pre-programme interview was performed to assess needs and to know the topics participants would like to see included in the programme; the following themes were mentioned: medical information and education about genetics, heredity and its implications; prevention and available prophylactic treatments; coping strategies to increase well-being; and psychological support for family members.

### 4.2.2. Recruitment

Participants were selected at the Hereditary Tumours Consultation of the Centre of Medical Genetics and Human Reproduction of the University Hospital of Coimbra, Portugal. Ethics approval was granted by the mentioned institution. Recruitment criteria included families in which at least one member of a family had tested positive for deleterious CRC's mutations, and living at a reasonable driving distance (less than one hour) from Coimbra. In this study all family members were eligible to participate in the group, whether or not they had undergone genetic testing, and whether or not they were mutation carriers. This was because genetic risk is a family issue that emotionally affects all family members, either related or not by consanguinity bonds (McDaniel, 2005; Sobel and Cowan, 2003). Exclusion criteria included children with age under 12, persons with psychiatric diagnosis, or disabilities capable of interfere with verbal expression. One family initially appointed for the programme renounced to participate before its beginning, due to the geographical dispersion of its members (especially siblings). Drop out rate in this study was 20% (one family).

For the recruitment process, we searched at the service data base for eligible participants. Phone contacts were made briefly outlining the study and inviting them to take part through a multiple family discussion group, while mentioning a subsequent phone contact to address availability to participate in this study. Such phone contact was made approximately two weeks later, asking permission to sent confirmation letters. Afterwards, letters were sent to those who agreed to participate, where the previous information was given in order to be accessible to other family members, and attaching an informed consent form (requiring its deliverance at the first session); for participants under 18 years old informed consent was authorized by one of the parents. A third

phone contact was made to deal with practical issues concerning the scheduling for the sessions (all were scheduled ahead), including a previous family interview. Finally, family interviews with the potential participants were made (mostly at the participants home or at the hospital) to collect demographical and psychosocial data, and to assess needs.

### 4.2.3. Participants

One group was set up, involving 4 families and 19 people (table 4.1.). Participant families included 3 to 6 members. In all the families participants were linked by blood kinship (mothers, brothers, sons, daughters, aunts and uncles) or relational ties (couples, girlfriends, brother-in-law and mother-in-law).

**Table 4.1. Participant's demographics, illness features and family history**

| **Participants** | ***n* = 19** |
|---|---|
| Mean age | 33.5 (14 – 56) |
| Sex | 13 females, 6 males |
| Race | 19 Caucasians |
| Marital status | |
| Married | 11 |
| Single (Dating/In a relationship) | 6 (4) |
| Widow | 2 |
| Mean years of education | 9.2 (4 - 17) |
| Mean number of months since last genetic testing | 30.5 (2 – 60) |
| Type of CRC susceptibility (in families) | 2 HNPCC, 2 FAP |
| Genetic risk status | 9 CRC mutation carriers |
| Personal history of cancer | 4 affected (1 symptomatic) |
| Mean number of CRCs in the family | 4.75 (3 -6) |
| Mean number of at-risk individuals in the family | 3.25 (2-6) |
| Mean cancer-related deaths in the family | 2.25 (1 - 3) |
| Number of participant families | 4 |

### 4.2.4. Procedure

A multifamily discussion group (Gonzalez *et al*., 1989; Steinglass, 1998) was performed, comprising four semi-structured sessions involving a skills-acquisition component and a didactic element (Table 4.2.). The group integrates a supportive rather then therapeutic orientation. The acquisition of practical and informational skills was emphasised aiming to overcome day-to-day problems, while engaging in group discussions on themes concerning family adjustment to genetic illness and heightened cancer susceptibility. Sessions were co-facilitated by two psychologists with training in medical family therapy. Facilitators supported participants in normalising feelings, and

reinforcing families' competencies and resources; circular questioning (Penn, 1982) was often used to mediate interactions between the group participants and to allow engagement in reciprocal comments. A geneticist was present in one session to address genetics education issues.

**Table 4.2. Programme description**

| Session | Format/Segment | Contents |
|---|---|---|
| 1 | Presentations | Presentations. |
| | Support | Impact of increased genetic risk in the family. |
| 2 | Education | Medical information. |
| | | Community based resources. |
| 3 | Support | Embracing family identity |
| 4 | Support | Problem-solving techniques. |
| | Ending | Stress management strategies. |

Each session was expected to last 90 minutes, although duration invariably extended to 120 minutes. All sessions were videotaped with the permission of all participants. The sessions were scheduled at the participants' convenience at weekends (Saturdays, 10:30am-12:00pm), and were entirely free of charge for participants. The programme comprised two components: i) education, aiming at providing up-to-date medical information about CRC's mutations, prophylactic treatment options, and community based resources; and ii) support, including disclosure and family reactions to testing and to cancer susceptibility status, emotional reactions and coping strategies, and family identity maintenance. Each session had flexibility to integrate themes that spontaneously emerged from the participants.

#### *4.2.4.1. First session*

This session aimed to explore the impact of genetic risk awareness in the family. Initially, the need to maintain confidentiality among participants was underlined, and permission to videotape obtained. Informed consent forms were collected. After presentations of both participants and facilitators (who also introduced the programme format), participants were gathered by family and encouraged to share the negative and positive aspects that had emerged in the family context after the genetic diagnosis were known, or after attending genetic counselling. It was emphasised that such experience is often perceived as alarming; however, thinking about the family functioning opens the possibility of recognising other significant aspects of such experience. Next, families were invited to think how they could overcome the negative aspects. Conclusions were shared among all participants. Finally, a home task was requested: each family was asked to gather and

talk about their doubts and concerns of their genetic risk status, in order to discuss these with the doctor at the following session.

#### *4.2.4.2. Second session*

This session provided information about the medical aspects of CRC's aiming to dissipate doubts and allow a better genetic illness management. A doctor (geneticist, third author) was present providing the most recent medical information on HNPCC and FAP testing, treatment options and related concerns, as well as answering participant's questions and doubts. The following themes were addressed: genetic and hereditary illness significance, mechanisms of hereditary transmission of the genetic susceptibility, and prophylactic treatment options for genetic risk management. Community based resources were also made available, such as Internet sites and patients' organisations' forums. Finally, participants discussed their doubts with the doctor.

#### *4.2.4.3. Third session*

The main goal of this session was to help families keep a sense of continuity in their own history (past – impact of genetic risk in the family life; present – current challenges; and future – upcoming projects). The session started with a brief explanation about how a crisis like the threat of cancer genetic risk for or its onset may affect families, creating a gap between the family's past identity and the frightening present, and putting the future on hold. Aiming to explore how genetic risk shaped the sense of family identity, facilitators invited families to gather *by family* and to think about their values before, during, and after the disclosure of genetic test results, asking them about the main features of family functioning in each period.

#### *4.2.4.4. Fourth session*

The last session aimed at helping the participants develop stress-management strategies, since high levels of stress and anxiety are commonly reported amongst genetic illness patients. Two types of coping strategies were addressed: problem solving and relaxation. Concerning the first strategy, facilitators stated the universal nature of experiencing heightened anxiety when facing the challenges of a (potential) life-threatening illness, and then asked each family to share some of their current problems on this aspect. Families have chosen a problem solving exercise based on decision-making for prophylactic surgery. The exercise was developed according to the following rationale: identifying the problem; naming it; sharing points of view; finding possible solutions; deciding on one alternative; carrying out that decision; monitoring to evaluate its effectiveness (McDaniel *et al.*, 2006). As for the relaxation, the normative value of stress was underlined through examples that showed how stress becomes "natural" when facing uncertainty circumstances.

Cognitive and progressive muscle relaxation training were introduced and performed as an additional resource to the strategies they already use to manage stress. Multimedia equipment was used to enhance the relaxation process.

### 4.2.5. Evaluation

The programme was evaluated through a focus group interview (Piercy and Hertlein, 2005), a method often used as needs assessment and programmes' evaluation. Participants' views were collected as a participatory process to identify the individual and family impacts. The purpose was to address the programme's applicability through the users' point of view, considering its potential use as a complementary tool for supporting families in cancer genetic counselling services. This interview method was chosen (Piercy and Hertlein, 2005) because: it is pertinent for exploratory approaches, since it is a rich method of revealing experiences and perceptions; it is appropriate for sensitive topics, which is the case of an intervention addressing genetic illness experiences, as it provides a safe environment for participants to share their understanding and insights in an interactive manner. Although focus-groups present several limitations, particularly the limited significance of its results, its small group approach may produce more elaborated accounts than those generated in individual interviews (Wilkinson, 2004).

The focus group took place one month after the last session, and was conducted by the programme facilitators. All participants agreed to cooperate. The interview lasted approximately 120 minutes in length, and videotaped under consent. The interview was applied according to the guidelines for conducting focus groups (Wilkinson, 2004), comprising the following topics: the functional aspects, contents and activities of the programme; individual and family benefits; suggestions to increase the sense of well-being.

### 4.2.6. Data analysis

The focus group interview was fully transcribed and analysed through a grounded theory design (Glaser and Strauss, 1967). Grounded theory is described as both a systematic and rigorous method, allowing participants to be the experts in describing their own experience (Glaser and Strauss, 1967; McAllister, 2001). Qualitative content thematic analysis was used to analyse the interviews (Patton, 1990). Open coding was used to summarize content and representative statements from recurring themes; constant comparison between the emerging themes was carried out by the first author and an independent researcher, to enhance reliability (Strauss and Corbin, 1998). Successive refinement through repeatedly reviewing the transcripts was made until consensus was reached. Quotes from the raw data were included as descriptive summaries illustrating the emerging themes.

### 4.3. RESULTS

We will report data from the activities that have been included in the programme sessions and from the evaluation focus group with the participant families. All participants' names were changed for confidentiality purposes.

#### 4.3.1. Data from the sessions

In the first session, families highlighted as positive aspects of their (potential) genetic susceptibility, the strengthening of emotional and relational ties among relatives, and the possibility genetic counselling gave them to inform other family members of their potential increased genetic risk (chances from prevention). The negative aspects included the uncertainty, fear and anxiety while awaiting for genetic test results, the physical implications of surveillance procedures (colonoscopy) and prophylactic treatments (colectomy), and the postpone of personal and family plans. To overcome the negative aspects, families stressed the importance of having access to medical education, of promoting family support, and of focusing on the positive and more relaxing aspects of life.

Second session gave participants the opportunity to discuss their doubts with the doctor; the more recurring queries were: medical surveillance procedures, reproductive and childbearing issues, latest technological advances in genetics, informative resources and preventive measures for descendants.

In the third session, when asked to think about their values before, during, and after the genetic positive test result, families' stated that their values remain the same across time, although the involvement in genetic counselling and the *arrival* of testing results had increased solidarity and unity among family members, including extended family and significant others.

#### 4.3.2. Data from the focus group

Participants considered that all practical aspects of the programme were adequate (number of sessions, duration and frequency), preventing dispersion without being a burden for personal and family life. Nevertheless, it was suggested that sessions' should be extended by one more hour with a 15 minutes pause (although keeping the four session format), in order to engage participants in depth discussion:

> *"I think the program was well planned; maybe it would be better if sessions could last one more hour, because that would give everyone a chance to equally express their point of view without time constraints; sometimes we had to limit what we wanted to say to give other participants a chance to speak, and some issues stayed pendent (…) and during*

> *the break, we could perhaps chat informally and get to know each other better"*. [Jorge, male, 24 years old]

The multiple family discussion group format emerged as one of the major benefits of their participation, as it allowed a supportive atmosphere which encouraged the sharing of personal and family experiences:

> *"I had no expectations, because I never had the chance to talk about this subject with anyone (…) in the end I enjoyed sharing and listening to the stories of people who were facing the same difficulties"*. [Maria, female, 39 years old]

The group experience of sharing also enabled emotional disclosure. One participant, who had recently been informed about his and other relatives' increased genetic susceptibility, mentioned the group as a useful place to disclose "difficult" feelings, while others stressed the benefits of release tension through mutual discursive engagement:

> *"At the beginning* [first session] *I was very upset about everything… I was upset with the doctors, I had no hope in genetic counselling…This group gave me the chance to calm down, to carefully listen to what the doctor was saying, to make questions, to listen to other people's doubts, and to learn. I feel more relieved now"*. [Vasco, male, 33 years old]
>
> *"These sessions allow us to better process all the things we were told, and also to release anger…I think it's very useful, actually, to talk about it"*. [Sofia, female, 29 years old]

Some long-term at-risk participants mentioned the opportunity of being "a role model" in helping other families cope with genetic disease across time:

> *"This group gave me the opportunity to share my experience after years of suffering, fear, and doubt, and to transform that into something valuable and helpful to other people who are now facing the same problems. I feel I was a model for some of them"*. [Fernando, male, 50 years old]

Participants also mentioned the opportunity to learn from other people's stories, while being given the chance to learn about "*other ways of facing this situation*":

> *"Here we could talk, listen, learn, and it also enabled us to help each other overcome difficulties. It's not easy to talk about these things and find someone with the same problems… "*. [Maria, female, 39 years old]

The educational session was viewed as key moment for an effective integration of medical information, helping to create a sense of control over health management:

> *"The session with the doctor was very important because it allowed us to better understand the illness and the genetics (…) these things are complicated and we only want to know what things we can do to control risk and manage our health. Here we had time to listen*

> *and to ask properly, because we had talked about our questions and doubts ahead…”*. [Jorge, male, 24 years old]

One participant had resumed her positive view about the programme as:

> “*(…) having a logical sequence; first we knew each other, then we shared our illness experiences and how our family managed to adapt to it across time, we were informed, and finally we could relax! These sessions helped me to set things up, to make sense of everything we’d been through*”. [Joana, female, 17 years old]

The collaborative approach and proximity adopted by the facilitators was stated as an important factor for the group functioning:

> *“Facilitators helped us feel comfortable in front of other families as they were careful and attentive at the same time; for instance, I’d liked the way they kept in touch with us when scheduling the sessions, and during the week, before the sessions…it showed interest”* [Helena, female, 51 years old]
>
> *“I think their posture was great because they adopted an informal role, almost sharing their points of view with us instead of actually instructing us…perhaps if they had assumed a more formal attitude we had felt more inhibited”*. [Hugo, male, 23 years old]

Some participants said that the relaxation techniques did not worked for them (“*I couldn’t relax because I wasn’t able to concentrate on your words*”; “*The beach theme as a calm inducing image didn’t work for me at all*”), although recognizing the pertinence of the theme.

### 4.4. DISCUSSION

This programme takes an intervention-based, pragmatic approach to psychosocial genetics, combining educational and supportive services with a family focus. It was designed to assist them in coping with the persistent challenges of increased genetic risk for CRC during the post-test period. This qualitative exploratory study adds for the literature as a family-centred intervention programme designed for helping cancer susceptibility families as an adjunct service for the genetic counselling psychosocial delivery.

Results suggest that the programme is well-structured regarding its duration, contents and methodologies, and that it meets the families’ psychosocial adjustment needs to face a genetically linked condition. Participants’ suggestions concerning the lengthening of the sessions may be discussed in the light of the sample characteristics, such as: i) the existence of a significant temporal hiatus since participants’ attended their last genetic counselling session (30 months, in average); and, ii) the lack of any type of psychosocial support obtained meanwhile. As time management is an important feature to moderate group interventions, facilitators tried to balance the flow of dialogues with a reasonable degree of rigour, in order to accomplish the sessions’

agenda and extent. Despite there were no objective indications of increased stress in participants due to time constraints, in some occasions it was difficult for the facilitators to move the discussions away of the mutual generated stories around genetic illness, as participants' had much to say about its effects on themselves and their families. If more space had been given for those interactions, sessions would invariably be lengthened, as further and in depth discussions may have occurred. In addition, those individuals with more verbal resources would arguably lead, preventing others to fully express themselves.

Concerning the psychosocial impact of the programme, we argue that the group's heterogeneity on the illness adaptation phase (Rolland and Williams, 2005) emerged as a benefit for participants, helping them to better envisage the chronic process inherent to the genetic condition across time. The opportunity for participants to listen to other individuals and families who had previously experienced similar circumstances, and, in some cases, to function as a role model for those who have recently experienced genetic counselling, helped to enact a mutual, sharing atmosphere which allowed normalising feelings about genetic risk. Among the group participants it was possible to acknowledge the intersection of biological and relational ties in a community environment, as extended family units derived from the index patient came to sessions. Moreover, the group context seemed to facilitate bonding and extra-familial networks, significant features for relieving feelings of isolation and the psychosocial burden at both personal and family levels through emotional expressiveness and mutual support. Aspects commonly described in other studies concerning heightened genetic risk families did not emerged in this study, such as issues of loss, grief and mourning (Fanos and Puck, 2001), which perhaps could have emerged in a larger sample size. It may have been possible that participants' felt inhibited to talk about difficult emotions in a group environment.

Notwithstanding, this intervention allowed not only mutual support among participants, but also had stimulated families' conversations about cancer and genetics, a resilience factor for psychosocial adjustment in cancer risk and genetic test uptake (McCann *et al.*, 2009). By the reciprocal process of narrating their medical family history in their own terms, and listening to others' stories, families were allowed to share their stressful experiences, thus making sense of new and more purposeful meanings about living with a genetic condition. Such may promote the accommodation of their familial illness narratives to the available modern technological advances, enabling novel trajectories (Werner-Lin and Gardner, 2009). This programme also helped to create a sense of perceived control over one's health management by integrating an educational/informative component. To gather family members in order to collect doubts about medical issues and the subsequent presence of the clinician provided an opportunity to increase medical awareness, arguably useful considering decision-making purposes.

### 4.4.1. Implications for clinical practice

Participant's accounts reinforce the adequacy to incorporate family-based interventions in the scope of genetic counselling. Recommendations for the inclusion of family therapists have been envisioned for genetic counselling practice as genetic diseases have been recognized as familial diseases (Enupu 1997; McDaniel 2005). It is important to mention that a developmental approach when considering family interventions should prevail; a life-cycle perspective suggests individual and family development co-evolve through consecutive normative stages with anticipated psychosocial tasks and changes (McGoldrick and Carter, 1999). It is therefore relevant to acknowledge the timing of genetic illness specific demands in the intersection of individual and family development (Brouwer-Dudokde *et al.*, 2002; Steinglass, 1998; Street *et al.*, 2000). Genetic predisposition for CRC's, particularly for FAP, with its age of onset and the common need for early age surveillance or prophylactic procedures, may have a pervasive impact on psychosocial or psychosexual development, or in partnering and family planning, if coinciding with adolescence or early adulthood, respectively (Decruyenaere *et al.*, 1996; Werner-Lin, 2008). For this purpose, the integration of psychosocial genetics / family systems health professional in the genetics healthcare team would be relevant.

Informational and supportive multifamily forums performed regularly throughout the different stages of the genetic counselling process may contribute for public genetics literacy, and stimulate intrafamily communication about genetics, as well as a way to connect families and the genetic health care team in a collaborative fashion (Cohen *et al.*, 2009; McDaniel, 2005). Reports from one-day workshops (Gonzalez and Steinglass, 2002) or retreats in rural distant regions (McKinnon *et al.*, 2007) designed to intensively address psychoeducational contents in multifamily formats, may function as an alternative option for families who cannot attend the programme sessions due to financial burden or to geographical distance from the genetics centre. This programme, or an identical intervention comprising an adapted multifamily discussion group approach, may be suitable to replicate to other genetic illness domains.

### 4.4.2. Limitations and research perspectives

The decision of using the programme facilitators to carry out the evaluation focus group deserves a critical observation. Given that the principal researcher (first author) conducted all the investigation procedures and contacts with the participant families, the influence of a *halo effect* must be considered (Patton, 1990), so that researchers and participants were both motivated to perform in an exemplary fashion. This may have impacted findings, namely through an over-appreciation of the programme's impacts from the participants' point of view, and also by the evaluators' predispositions. On the other hand, the previously and time-extended participants-researchers

relationship might have produced a tension-free environment suitable to encourage honest reports. In line with second-order cybernetics' epistemology, it is our belief, however, that neutrality in qualitative enquiry means impartiality rather than distance, and that the observer cannot be excluded from the observed reality (Foerster, 1996). If outside researchers had assured the evaluation focus group for the sake of *objectivity*, the over-appreciation issue would not be excluded, and a top-down, "specialist" relationship with participants could occur or at least be perceived as such by the participants, possibly making them feel more anxious to the presence of external evaluation. In order to obviate such potential biases, more than one evaluation moment could perhaps enhance reliable outcomes.

As with many other exploratory studies, the sample was small and therefore not representative. Nevertheless, our goal it is not to make generalisable empirical claims but rather to use the data qualitatively to suggest ways for fruitful family-oriented support on genetic counselling. Furthermore, data collected from participant's accounts on their participation in this programme are suitable to include biases by factors that encourage families to take part in research, such as the lack of psychosocial support obtained meanwhile, the commitment to the alleviation of suffering of others with the illness, or the will to be engaged in scientific development.

Hence, further research should include a larger number of families and be carried out at other genetic counselling services to evaluate the applicability of this programme. Performing this programme at other phases of the genetic counselling timeline (Rolland and Williams, 2005) should be considered as well, namely in a pre-test context and immediately after knowing the genetic test results, adapting the sessions' contents accordingly to the psychosocial specificities of each phase. There is also the need to enhance our understanding of specific intra-familial dynamics throughout the genetic counselling experience, namely how communication of genetic information within families occur.

## 4.5. CONCLUSION

Overall, this programme provides a useful tool for follow-up psychosocial support for CRC susceptibility families, complementary to the more biomedical driven and person-centred protocol of genetic counselling. As adaptation to increased genetic risk is a multidimensional, ongoing and chronic process, there is a need for long-term follow-up support for personal and family adjustment to genetic illness in its complex psychosocial milieu. This programme has potential for supporting families in the scope of cancer genetic counselling.

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# CAPÍTULO III

## INCORPORAÇÃO DE APOIO PSICOSSOCIAL NO ACONSELHAMENTO ONCOGENÉTICO: PERSPECTIVAS DOS PROFISSIONAIS

# 5. ARE FAMILY-ORIENTED INTERVENTIONS IN PORTUGUESE GENETICS SERVICES A REMOTE POSSIBILITY? PROFESSIONALS' VIEWS ON A MULTIFAMILY INTERVENTION FOR CANCER SUSCEPTIBILITY FAMILIES[30]

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**ABSTRACT**

This article examines genetics healthcare professionals' opinions about a multifamily psychoeducational programme for hereditary cancer susceptibility families, implemented at a Portuguese genetics service. Their views on how a family-oriented approach is envisioned to be incorporated in Portuguese genetic counselling services are also reported. 6 focus groups and 3 individual interviews were undertaken comprising 30 professionals working in the provision of genetic counselling, and genetic counsellor trainees. Participants were given a page-summary describing the intervention, and asked to comment the strengths and limitations of the multifamily intervention. All interviews were fully transcribed and analysed using the constant comparison method. The qualitative analysis generated data comprising four thematic categories in relation to the professionals' views: i) usefulness of the programme; ii) programme' methodological and practical obstacles; iii) genetics services constraints; and, iv) suggestions for improving the programme and further family-oriented interventions. We reflect on the reported views examining the intervention, and on how current constraints of genetic services limit the provision of psychosocial support for cancer susceptibility families. These findings may sensitise stakeholders and policy makers for the need to deliver family-based services in cancer genetic counselling, with adequate planning and collaborative involvement of different professionals.

**Keywords:** cancer genetic counselling**;** family-centered services; intervention evaluation; multifamily discussion group; reliability of qualitative data.

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## 5.1. INTRODUCTION

Genetic counselling and testing are familial experiences that often confront individuals and family members with difficult medical management decisions, commonly involving complex ethical, legal and psychosocial issues. The need to develop family-oriented models of coping, adaptation and integration in inherited risk management has been stated as crucial to fully address the holistic needs of those seeking help from genetic counselling services (McDaniel 2005; Peters et al. 1999; Street et al. 2000).

Group interventions for individuals and their relatives with cancer susceptibility mutations are one of the most common ways to provide psychological support and educational information. Psychoeducational multifamily groups have been well established as family-focused interventions for chronic medical illnesses (Asen 2002). Albeit there is some debate concerning its effectiveness and moderators (Esplen 2011), such approaches have been stated as a useful tool to address the psychosocial interface between medical, individual and familial issues (Rolland and Williams 2005) because they typically include both patient and family members. Whilst it has been used for supporting individuals and their families facing several chronic illnesses, including cancer, research on multifamily groups are scarce or non-existent in the field of psychosocial genetics.

Literature is prolific in examples of group approaches for individuals at increased oncogenetic risk. Esplen et al. (2004) studied the use of a supportive-expressive group for *BRCA* mutation carriers comprising 8 weekly sessions followed by 4 monthly sessions. This intervention addressed the emotional impact associated with having a family history of breast/ovarian cancer and being at high risk status for developing cancer. Participants were found to have improvements in psychological functioning, by reducing depression and anxiety levels, and several women made decisions concerning prophylactic surgery during and after the intervention; additionally, some of these women also revealed a reduction on grief feelings from pre- to post-intervention measures (Esplen and Hunter 2002). Specific psychoeducational interventions for *BRCA* carriers comprising educational and psychosocial components are also well established. Wellisch et al. (1999) conducted a pilot study of a psychoeducational group intervention for high risk relatives of breast cancer patients, reporting a decrease of psychological burden in participants. Kash et al. (1995) reported a randomised controlled trial of a one-year group intervention, describing a reduced perception of risk and an increased adherence to screening behaviours and in knowledge about participants' high risk status. In a psychoeducational written intervention consisting of an educational and psychosocial pack, Appleton et al. (2004) also found post-intervention benefits in terms of diminished cancer worry and improved *BRCA* risk-related information. Karp et al. (1999) described a psychoeducational programme reporting prophylactic mastectomy issues in a group of

*BRCA* carriers, and Speice et al. (2002) conducted a six-session psychoeducational intervention focusing on family-related themes. All of these interventions, however, did not include family members besides the patient, who may also benefit from participation.

In this article, we report findings from an exploratory qualitative study with a sample of Portuguese genetics healthcare professionals on how they assess a multifamily psychoeducational programme for hereditary cancer susceptibility families. The programme design, implementation, and evaluation are described elsewhere (Mendes et al. 2010; 2011). Its aim was to assist at-risk families in coping with the ongoing demands of increased genetic risk for hereditary cancers during the post-test period, combining educational and supportive services with a family focus. Two multifamily groups were performed, involving 7 families (4+3) and 28 individuals (19+9) were involved including biological relatives and other family members; participants attended 4 weekly sessions. Sessions were co-facilitated by two psychologists trained in medical family therapy and with previous experience with multifamily groups. Participants evaluated the programme through focus groups, suggesting that the programme is well-structured regarding the duration, contents and methodologies, and that it generally responds to the families' needs of adjustment to genetic illness and its increased susceptibility by enhancing their well-being and coping resources.

In Portugal, cancer genetic counselling is available in oncological hospitals and integrated at major hospitals' oncology or genetics departments. The first generation of Portuguese genetic counsellors recently finished their training of a two-year professionalizing master course in genetic counselling, administered from the University of Porto. The course benefited for inter-European experts participation, and national-based institutional support for practical observation. Genetic counselling in Portugal is mainly assured by medical geneticists.

In this paper we present an additional study reporting genetics healthcare professionals' views on this multifamily programme, and on how they envisage the incorporation of family-oriented approaches in genetic counselling services. Our aim is to enhance credibility to the overall study by cross-checking participant families' and professionals' perspectives, and to strengthen the validity and reliability of data through triangulation of qualitative data sources (Patton 1990). We also sought to reflect on the current needs and constraints' Portuguese genetics services face for incorporating suitable psychosocial services for families. We describe our analysis of professionals' qualitative individual and focus group interviews on the usefulness and limitations of the intervention, and how they envisage its incorporation at Portuguese genetics services. Suggestions for further family-oriented themes and interventions in the scope of cancer risk counselling are also described.

## 5.2. METHODS

A qualitative study design was used, since the main emphasis was to explore people's experiences and perspectives in an area where little is known to guide research or practice (Glaser and Strauss 1967; McAllister 2001). Semi-structured individual and focus groups interviews were applied according the interviewees' convenience. Our initial purpose was to perform focus groups; however, 3 individual interviews were undertaken due to difficulties in finding a common schedule for the focus group. Focus groups were preferred over individual in-depth interviews because the group setting allows individuals to use the ideas of others as cues to fully elicit their own views, which may stimulate topics of discussion and therefore contribute to create a richer source of data (Piercey and Hartleim 2005). Although focus groups are described as a useful method for exploratory approaches, as they use the group dynamics to gain insights and generate ideas, and previous research has indicated its suitability in working with genetics professionals (McAllister et al. 2007; 2010), they also present limitations: confidentiality is not possible in group settings, and the group setting may be inhibiting to some participants (Piercey and Hartleim 2005).

A list of public institutions working in cancer genetics were identified using the Health General Directory (2004) and by key personnel. An email was sent to the directors of: 7 medical genetics departments of major and regional hospitals; and of 3 oncological hospitals. The email included an invitation to the genetics healthcare teams to take part in a focus group interview aiming to explore their views on a previously implemented multifamily intervention for at-risk cancer families (Mendes et al. 2010; 2011); professionals from different backgrounds were eligible to participate. From the 10 contacted institutions, 2 declined to participate; motives were not explored due to ethical reasons. The focus group interview guide (Table 5.1.) and a page-summary describing the intervention programme were sent to those who agreed to participate. Subsequent email contacts were established to arrange the interviews. One focus group gathered professionals from 2 institutions from the same city. Overall, 3 individual and 6 focus group interviews were conducted by the first author (AM) and a genetic counsellor from one of the genetics professionals' institution (MP), lasting approximately 1 hour. Further details about the intervention were provided at the interview.

**Table 5.1. Interview guide**

| Topics | Questions |
|---|---|
| Multifamily intervention in cancer risk counselling | *What general considerations do you want to make about the programme?* |
| | *What are its potentialities and its limitations?* |
| | *What kind of readjustments in its contents and structure do you suggest?* |
| | *How do you envisage the incorporation of this programme and other types of family support in Portuguese genetics services? What would be needed?* |

A total of 30 professionals working as part of genetics healthcare teams were interviewed, including 17 geneticists (from which 4 were interns), 2 oncologists, 1 obstetrician, 3 genetic nurses, 3 psychologists and 4 genetic counsellors' trainees (3 psychologists and 1 nurse) (Table 5.2.). Participants represented 8 institutions delivering cancer genetic counselling (4 major hospitals, 1 regional hospitals, 1 oncological hospital, and 2 genetics centres); the genetics centre is mainly devoted to genetic counselling for late onset neurological disorders.

Interviews were audio-taped with the participants' consent, fully transcribed and submitted to content analysis. Open coding, to summarize content and representative statements from recurring themes, and constant comparison between the emerging themes, were used (Patton 1990). The first author and an independent researcher performed successive coding refinement through repeatedly reading the transcripts, aiming to develop consensual content categories; categorization titles were given to similar themes and contents (Strauss and Corbin 1998).

**Table 5.2. Summary of focus group and individual interviews composition**

| **Focus Group** | **Professional background** | **Number of participants** ($n$= 30) |
|---|---|---|
| 1 | 6 geneticists (1 intern) | |
| | 1 genetic counsellor trainee (psychologist) | 7 |
| 2 | 1 geneticist | |
| | 2 nurses | |
| | 1 genetic counsellor trainee (psychologist) | 4 |
| 3 | 1 obstetrician | |
| | 2 oncologists | |
| | 1 nurse | 4 |
| 4 | 6 geneticists (3 interns) | |
| | 1 genetic counsellor trainee (psychologist) | 7 |
| 5 | 2 psychologists | 2 |
| 6 | 2 geneticists | |
| | 1 genetic counsellor trainee (nurse) | 3 |
| **Individual interview** | | |
| 1 | 1 psychologist | 1 |
| 2 | 1 geneticist | 1 |
| 3 | 1 geneticist | 1 |

## 5.3. RESULTS

The qualitative analysis generated data comprising four main themes, each comprising content categories, in relation to the professionals' views (Table 5.3.): i) usefulness of the programme; ii) programme's methodological and practical obstacles; iii) genetics services constraints; and iv) suggestions for improving the programme and further family-oriented interventions.

**Table 5.3. Summary of themes and quotes**

| Themes | Participant quotations |
|---|---|
| **1. Usefulness of the programme** | |
| Enhancement of well being | *"Support outside the biomedical scope, involving psychologists and social workers, are always useful because sometimes people need to cope with difficult decisions and these interventions help to relieve stress".* |
| | *"We need to look for the entire individual".* |
| Mutual support | *"People may feel bonded and close to others dealing with similar challenges, especially because we are talking of relatively rare diseases"* |
| | *"I believe this can be very therapeutic as in many cases these families feel isolated and stigmatised".* |
| **2. Programme's methodological and practical obstacles** | |
| Lack of quantitative outcomes | *"A pre- and post-test would be important to measure modifications in key outcomes, such as psychological adjustment or in information management"* |
| | *"The quality of the genetic counselling people had before will influence how intervention impact participants".* |
| Sampling and generalization | *"Participant families were probably the more adjusted, with more socio-economic conditions, and the more motivated".* |
| Recruitment and mobilisation | *"I believe that few families are available to be part of an intervention for a month, two hours every week (…) it is very intensive and impossible to universalize"* |
| | *"It's not eminently medical, so people will not consider it as absolutely necessary (…) most of these people are active, they work, and in many cases they fear problems at working places because the justifications".* |
| Group setting constraints | *"Some people may feel uncomfortable talking in front of others (…) it is impossible to assure confidentiality in a group setting".* |
| **3. Genetics services constraints** | |
| Scarcity of qualified human resources | *"A multidisciplinary team here [in the genetics service] would be vital, with psychologists working fulltime rather than being called from other services; we neither have time or training to do it".* |
| Physical barriers | *"We are facing other needs, such as physical space, available at the hospital".* |
| Funding | *"Usually this kind of interventions are developed in the scope of research projects with someone highly motivated, but afterwards things do not go further and people cannot benefit anymore once its over".* |

| **4. Suggestions for improving the programme and futher family-oriented interventions** | |
|---|---|
| Partnerships with community-based services | *"I conceive this type of intervention more in the scope of community- based services or patients representatives than at the hospital setting, which may dissuade participation from other family members"*<br>*"We should work together, but in a formal basis (…) referrals, for example: in many cases things work because we informally know other professionals who can provide some support!"*. |
| Integrating crucial issues as a complement to genetic counselling | *"Other more relevant issues should be integrated as a complement to genetic counselling: for example, understanding peoples' needs for communicating with relatives, help them in decision-making, or knowing their informational needs, so I can change my way of transmitting genetic information"*<br>*"It could be interesting to perform follow-ups to assess new concerns and relevant information"*. |
| Genetic counsellors | *"I am optimistic because we've started to produce genetic counsellors; they should be encouraged to integrate family-based concepts in genetic counselling training and practice"* |

**Table 5.3. Summary of themes and quotes** (cont.)

### 5.3.1. Usefulness of the programme

Throughout the group discussions, the vast majority of participants generally shared their positive views regarding the intervention. They pointed out its usefulness and acknowledged the need to help families' adjustment to their cancer susceptibility status in the scope of genetic counselling. Major perceived benefits regarding the programme relied on: i) enhancement of well-being; and ii) mutual support.

i) The intervention has the potential to alleviate the psychological burden that some families in this context often carry; it is also seen as an opportunity for non-biomedical support, contemplating a broader care for the individuals.

ii) The mutual support atmosphere posed by the group context, mostly stressed as a way to promote paths of successful coping through sharing experiences.

### 5.3.2. Programme's methodological and practical obstacles

Despite recognizing its potential usefulness, participants also mentioned the need of practical changes in the intervention in order to meet conditions for its consistent incorporation in genetics services. The mentioned obstacles were the following: i) lack of quantitative outcomes; ii) sampling and generalization; iii) recruitment and mobilisation; and iv) group setting constraints.

i) The lack of quantitative outcomes was consistently reported as a methodological limitation of robustness of this intervention. For example, it was argued that the

perceived quality of the delivered genetic counselling would influence the counselees' needs; addressing participants' previous informative knowledge before the intervention was then reported as necessary. It was presumed that even if participants were unhappy with the quality of the genetic counselling they attended, they would be likely to rate the intervention as useful anyhow.

ii) The sampling method was considered problematic by the participants, because the programme participants were not representative of the general Portuguese population, as recruitment occurred in a specific area covered by the genetics service where the study was carried out (centre region of Portugal); generalizable assumptions about its validity and sustainability were then described as limited.

iii) Some professionals reported the intensive nature of the intervention programme (4 weekly 90 minute sessions) as inadequate and potentially burdensome for the majority of families, limiting recruitment particularly for those coming from distant and rural regions.

iv) Group setting constraints were also mentioned as potentially limiting people's involvement, namely because of privacy issues.

### 5.3.3. Genetics services constraints

In order to incorporate this programme or to provide family-oriented interventions in genetics services, professionals' reported structural constraints centred on: i) scarcity of qualified human resources, ii) physical barriers; and iii) limited funding.

i) Limited human resources in genetics healthcare teams prevent the delivery of appropriate psychosocial interventions. The lack of a multidisciplinary team including genetic counsellors, psychologists and social workers was consistently described as an obstacle for implementing this and other psychosocial-oriented approaches. In some cases teams do not integrate full-time practitioners; professionals commonly assist other services at the hospital, generating work overload and affecting directly the availability of specific psychosocial assessment and interventions.

ii) Confined space was described as a specific limitation in some genetics departments. Performing group interventions with several families would hardly be possible in some hospitals.

iii) Lack of funding is the professionals' attribution for the above mentioned constraints. With available funds, continuity between time-limited research projects and its subsequent incorporation in the service delivery, if justifiable, would be possible.

### 5.3.4. Suggestions for improving the programme and further family-oriented interventions

Professionals shared some suggestions to aid the programme applicability:

i) The development of partnerships between genetics clinics and community-based services (such as patients and family representatives' organisations, primary healthcare practitioners, in particular family physicians, and health care centres) was highlighted as a way to enhance the feasibility of this intervention. Namely, establishing referral channels with those services in a local/regional basis will possibly help people from distant, rural areas to participate, as well as those with limited financial resources.

ii) More pertinent topics for the genetic counselling process should be considered for integrating the programme's contents, such as: helping counselees to better communicate with relatives about genetic risk and testing, specific aids to assist decision-making, or clinician-patient risk communication. Also, the inclusion of periodic follow-ups was mentioned as a way to provide ongoing support for families and to keep them linked to genetics services for reporting pertinent new information concerning risk management.

iii) In addition, the prospect of integrating newly trained genetic counsellors in Portugal in genetics services is seen as a major potential contribution to enhance a family-oriented approach to genetic counselling.

## 5.4. DISCUSSION

Similarly to other psychoeducational interventions for oncogenetic high-risk individuals, our programme addressed the implications of increased cancer susceptibility on family relationships, facilitated coping skills and provided medical information about risk. However, as the new genetics has reconfigured the family unit as the patient when inherited conditions are diagnosed, our intervention included both the patient and other *non-ill* family members, differently from the majority of known psychoeducational interventions (Esplen et al. 2004; Karp et al. 1999; Kash et al., 1995; Speice et al. 2002; Wellisch et al. 1999). Although support groups for families and caregivers are common, groups joining patients and their relatives remain unusual. Moreover, while other interventions generally took a quantitative evaluation from participants, our study qualitatively analysed genetic health professionals' views on the programme and how they envisage the incorporation of a family-orientated approach into genetic counselling services.

Since participant families' views on the adequacy of the programme concerning its structure and contents were previously addressed (Mendes et al. 2010; 2011), in this study we pursued to refine the programme's evaluation through an inter-professional reflective process. To our knowledge,

this is the first study to report the evaluation of a psychosocial intervention from the healthcare professionals' perspective. In spite of its limitations, this study represents a participatory strategy for engaging providers of genetics services with a family-focused perspective. It also contributes for providers to be reflective about their work, a useful professional tool since a shift in models of service delivery has been discussed (Battista et al. 2011; Wham et al. 2010). As Portugal, like many other countries, lacks an integrated plan for the provision of oncogenetic services, feedback from professionals represents a key aspect when considering the development of supportive interventions for those at increased risk and their families. Furthermore, it is a valuable input in terms of the formative and process components of programme evaluation, namely in negotiation and planning its development and implementation (Metcalfe et al. 2008).

Genetics healthcare professionals working in the provision of cancer genetic counselling, with different training and experiences, shared their views whether the intervention provides a feasible and useful tool to help at-risk families. While professionals reflected on how to enhance the programme structure and effectiveness for its incorporation in genetics services, current constraints affecting the delivery of appropriate psychosocial services in the scope of cancer genetic counselling were also highlighted, lending further insight to the barriers families experience when attending genetics services. Such limitations also stress the need for integrating adequately trained healthcare professionals in cancer genetics services. Therefore, this study provides an exploratory account on the current challenges genetics services in Portugal are facing in order to provide integrative genetic counselling services for at-risk cancer individuals and their families.

### 5.4.1. Learning from our experience

The positive endorsement from participating families (Mendes et al. 2010; 2011) parallels the professionals' views on the usefulness of the programme. However, despite the growing awareness of the need for a family-focused approach in the scope of increased genetic susceptibility, some barriers may contribute to explain the paucity of time-extended group interventions. Aspects such as recruitment restrictions to a mainly urban population, the need to regularly coordinate scheduling demands with participant families, financial costs and the lack of staff trained in family systems are among some obstacles for the universalization of these interventions. Therefore, a condensed version of the programme in a one-day multifamily workshop may overcome such difficulties, besides its potential for cost-effectiveness and for allowing a more realistic dissemination of psychosocial care in distant regions from genetics centres. Such model was performed for chronic medical illnesses (Steinglass et al. 2011) as well as in the scope of genetic risk (McKinnon et al. 2008), comprising components of lectures and small group discussions around specific themes (medical updates on cancer risk management, family communication and

genetic testing or spouse/partner issues). However, by contemplating just one single "moment", this model may result as a scarce effort for embracing the ongoing psychosocial demands of those facing oncogenetic risk and their families. In order to potentiate a broader impact, follow-up "booster" sessions after an intensive one-day workshop experience have been argued by Steinglass et al. (2011) and already used by Esplen et al. (2004). Psychoeducational written material including information on scientific and psychosocial aspects of familial risk for cancer may also be distributed, as described in the study of Appleton et al. (2004).

Methodological limitations pointed out by professionals need to be acknowledged for the programme's applicability purposes. The absence of summative evaluation components weakens the validity of the intervention. Collecting baseline data for comparison with post-intervention short- and long-term information and psychosocial effects, or, better still, using randomised groups, are core issues that may well be taken into account in future developments. As with many exploratory studies, the sample was small and purposive, and therefore not representative. Our aim, however, was not to make generalizable empirical claims about a wider population, but rather to use the data qualitatively to address how participants evaluate the programme's structural and practical aspects and the perceived benefits in their lives. Moreover, professionals' reports about the programme's lack of objective outcomes and sampling representativeness evidence the assumption that research in genetics should mainly assume quantitative purposes. This assertion may perhaps be fuelled by the participants' dominant medical sciences background, greatly rooted in a predominantly quantitative, evidence-based orientation in the field of enquiry.

The focus group format prompted professionals to discuss about the most common perceived needs of their counselees, and to envisage the inclusion of other ways to support families in oncogenetic counselling. Alternative and more critical themes were identified as potentially adequate to be included as contents in this programme, performed in a complementary fashion to genetic counselling, such as decision-making for risk reduction options, or facilities to enhance families' communication skills to disseminate genetic risk information to other potentially at-risk relatives. Individuals and families attending oncogenetic services need psychosocial support in other ways besides multifamily interventions. As literature states multifamily groups are primarily effective for cohesive families (Campbell 2003), clinics must provide diverse psychosocial tools for supporting not only less cohesive families, but patients (and their families) with other characteristics as well.

### 5.4.2. Family-oriented interventions in genetics services: a remote possibility?

The concepts of family system theory and recommendations for the inclusion of family therapy trained professionals have been envisioned for genetic counselling practice as genetic diseases have been recognized as familial diseases (Eunupu 1997; McDaniel 2005). Although the incorporation

of family dimensions as an intrinsic part of the genetic counselling delivery has been stated as an important feature in cancer genetic counselling (Kenen et al. 2003; Street et al. 2000), professional's accounts evidence several limitations to such endeavour, at least in the Portuguese scenario. One of the issues discussed at length in the interviews was the lack of qualified human resources, notably those more markedly rooted in a psychosocial orientation (psychologists, social workers or family therapists), and in some cases of physical conditions at the genetics services, as a way to explain the scarce provision of psychosocial support for families. In some cases, such support is given by professionals from other internal, and even external, services. One might consider that these constraints only represent the visible part of these unmet needs.

A wider approach to genetics services, and specifically considering the genetic counselling protocol, may represent an initial step towards a more psychosocial sensitive delivery, suitable to include family support. Community-based facilities, such as patient or family representative's organisations, were stated by some professionals as a more appropriate setting to deliver these interventions instead of the hospital. Community services may indeed perform an important role as an adjunct facility of genetics services, linking tertiary and primary assistance, although demanding a careful management of ethical and co-ordination issues. The role of primary care physicians should also be considered. Lack of training and of confidence of these professionals to carry out medical genetics tasks has been stated (Nippert et al. 2011). Given the amount of patients primary care professionals refer for oncogenetic services, training in specific issues about genetics, namely in how to make appropriate referrals for cancer risk counseling, may indeed improve the service.

Such endeavours require a significant policy shift in the current provision of cancer genetic counselling in Portugal, and, perhaps more importantly, in its planning. Theoretical and practical education is required in order to gain understanding and skills in implementing specific interventions (Jacobsen 2009); besides the harmonization of practices and professional recognition, the training of (non-) genetics healthcare professionals is currently one of the greatest challenges for genetic counselling across many countries (Skirton et al. 2010).

Yet Portugal is among the group of European countries having specific legal provisions on genetic counselling practice (EuroGentest 2008), the outlined constraints emerge as encapsulated by the inexistent tradition of psychosocial practice in genetics settings, which may in some extent perpetuate the assumption that family support and genetics are incompatible matters. As genetics service providers receive adequate training in psychosocial issues, healthcare professionals' levels of confidence in dealing with more family-oriented tasks may increase and thus be suitable for inclusion as a feature of patient care. The inclusion of newly trained genetic counsellors in the mainstay of cancer genetics services may represent an opportunity for the provision of more psychosocial-oriented interventions, namely by assisting patients and their families throughout the

genetic counselling protocol, helping with psychosocial assessments, and establishing links between the genetics clinic, primary care and community-based resources.

### 5.4.3. Implications for cancer genetic counselling

The need to provide adequate psychosocial accompaniment was envisioned as a cornerstone for services based on mutation-based predictive technology (Stiefel et al. 1997). Kessler's teaching and counselling models are commonly touted as the primary approaches for genetic counseling practice (Kessler 1997). Such models were inherited from different professional backgrounds, posing genetic counseling as a frontier discipline between the realms of biomedical and psychosocial healthcare (Lewis 2002). In fact, genetic counseling can include both approaches through a psychoeducational focus adapted to different contexts (as oncogenetics), to consultands' idiosyncrasies and to local services peculiarities (Biesecker 2001).

The inclusion of a pre-counselling psychosocial assessment, performed by adequately trained professionals, may enhance the genetic counselling process, as information needs, personal and family medical history, risk perception and beliefs and family communication patterns and resources may be explored. This is suitable to alleviate the counselling agenda and to focus communication within subsequent sessions. Furthermore, in accordance with previously assessed needs, tailored family-focused psychoeducational modules delivered at key points of the genetic counselling protocol may constitute a suitable way for providing pre- and post-testing, ongoing support for consultands and families.

## 5.5. CONCLUSION

Findings from this study call for collaborative work amongst different healthcare providers working at primary, secondary and tertiary levels as a way to improve reliability of a multifamily intervention, and, more broadly, to enhance a family-oriented focus in cancer genetic counselling. This study provided an exploratory cross-checking to the examined programme, and to map needs for developing further and more adequate family-based interventions for cancer susceptibility families. Validated and tailored interventions may improve the quality of genetic counselling services, arguably contributing to counselees' and their families' empowerment, a qualitative outcome for genetics services that is currently under refinement (McAllister et al. 2010). As the first generation of Portuguese genetic counsellors is completing their training, these findings may sensitise stakeholders and policy makers for the need to integrate psychosocial support for families in the scope of cancer genetic counselling, an effort that will require adequate planning and collaborative involvement of different genetics healthcare professionals.

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# 6. CHALLENGES FOR CANCER GENETIC COUNSELLING: A QUALITATIVE STUDY IN PORTUGUESE ONCOGENETIC SERVICES[31]

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## ABSTRACT

In some countries, oncogenetic counseling services are still not provided by specifically trained professionals. Psychosocial support is only partially made available for individuals and families throughout genetic counseling protocols. The purpose of this study was to examine the current practice of oncogenetic counseling through the professionals' views and to ascertain the needs for the provision of psychosocial support in cancer genetics services. A qualitative study was designed; semistructured focus groups and individual interviews were performed involving 30 professionals from Portuguese healthcare institutions where oncogenetic counseling is offered. Current practice, unmet service needs and issues for improving practice were the major themes identified in participants' perceptions. Findings suggest: professionals' practice is aligned with the teaching model, with a nondirective focus; a scarce workforce of adequately trained psychosocial professionals, aggravated by other structural and organizational constraints are serious drawbacks on psychosocial delivery; multidisciplinary teams working in genetics were stated as priority, along with genetics education for healthcare professionals in primary care. Implications for practice and policy are discussed. Cancer genetics healthcare needs an adequate training and organization in order to integrate collaborative standards of care and functional forms of access for patients.

**Keywords:** oncogenetics; genetic counselling; cancer genetics services; qualitative study.

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## 6.1. INTRODUCTION

The most used definition for genetic counseling was proposed in 1975 by The American Society of Human Genetics (ASHG 1975). Although some revisions has been done since then, genetic counseling is still being considered as a communication process which deals with the human problems associated with the occurrence, or the risk of an occurrence, of a genetic disorder in the family (Resta 2006; Resta et al. 2006; Skirton et al. 2010a).

Cancer risk counseling has grown rapidly in recent years to become a major area of specialisation within genetic counseling. It was initially adapted from traditional genetic counseling protocols for hereditary adult onset disorders, namely Huntington disease, although differing significantly from the latter due to the availability of prophylactic medical interventions (Schneider 2002). Commonly used guidelines for cancer genetic counseling includes collection of personal and family medical history, psychosocial assessment, cancer risk assessment, pre-test counseling and informed consent for susceptibility testing, and disclosure of results; surveillance, preventive screening, treatment and follow-up sessions are also recommended (Trepanier et al. 2004).

The present study aims to examine the current practice of cancer genetic counseling through a sample of Portuguese genetics healthcare professionals. This study also sought to ascertain the needs for the provision of psychosocial support in cancer genetics services.

### 6.1.1. Models of practice

Historically, the genetic counseling practice has relied upon models from medicine, education, and mental health (McCarthy-Veach et al. 2003). In fact, genetic counseling is a classic example of the change in the doctor-patient relationship where the premise is that information cannot be owned by the professional, as the individual has rights of access (Evans 2006). Moreover, genetic counseling is often viewed as a frontier discipline between more “traditional” biomedical health care fields and a more psychotherapeutic and psychosocial one (Lewis 2002).

Kessler (1997) described two major models commonly touted as the primary professional approaches of genetic counseling practice: a teaching model, adapted from academic medicine, and the counseling model, adapted from mental health professions. The first assumes education of consultands as its major goal, based on the perception that clients come to genetic counseling seeking for information. The counseling model, in contrast, assumes as the primary goal of genetic counseling the understanding of the consultands’ complex motivations for attending the clinic, support client’s ability to cope and gain control over events involving genetic risk or conditions. Follows a person-centred approach, where conveying information is central, but it is not the primary goal. In parallel to these models, the concept of nondirectiveness has been linked to genetic counseling definition as a central tenet of practice (Weil, 2003).

Inherited from Rogers's client-centered approach, nondirective counseling aims at promoting the autonomy and self-directness of the client, and is closely linked to counseling approaches and techniques, as well as to professionals' identity. Since closely linked to a set of skills from the counselor, nondirectiveness has raised problematic issues regarding its imprecise definition and its ethical insufficiency, besides its *vulnerability* to the counselor's attitudes and institutional constraints (Weil, 2003). This is especially relevant in genetic counseling for common diseases with both genetic and environmental components (as for the case of cancer genetics), namely regarding issues as decision making for genetic testing and prophylactic interventions. In that sense, the need to bring a psychosocial awareness as the fundamental role of the genetic counseling work is emphasized (Weil, 2003).

### 6.1.2. Establishing standards of practice

Alternative approaches for genetic counseling practice are beginning to be discussed but little is known about how they are articulated in to routine practice (McCarthy Veach et al. 2007). Although genetic counseling is a well-recognized profession in countries as USA and UK, the development of practice and standards are still beginning in some others countries (McCarthy Veach et al. 2007; Skirton et al. 2010a). There is a growing need for the genetic counseling profession to develop models and standards of practice, defined by the practices of genetic counselors (Resta 2006; Skirton et al. 2010a). In Europe, harmonizing professionals' education across many different countries and healthcare settings has been stated not only as a valuable foundational work for establishing the profession of genetic counselor, but as a major contribution to ensure appropriate quality of genetic counseling services as well. Contents of education and training of genetic counselors are being established, as well as the set of roles, practice, and core competences at European level (Skirton et al. 2010a).

### 6.1.3. The Portuguese scenario

Portugal, with around 10 million inhabitants, has 6 Medical Genetics services: 2 in Porto (littoral North), 1 in interior North, 1 in Coimbra (Centre) and 2 in Lisbon (capital) (Health General Directory 2004). In 1999 Medical Genetics was recognized in Portugal as a specialty. Ten years later the first generation of genetic counselors started a formal training course as defined following core competences of ESHG for genetic counseling (Health General Directory 2004; Skirton et al. 2010b). This is a two-years professionalizing master course, its main objective is to train professional counselors, to join multidisciplinary teams at medical genetic services. Some teachers and students of the master course are already part of the European Network of Genetic Counselors

(ENGNC) and Community Genetics Network but any National Association has been created. To the present moment 53 medical geneticist has been graduated but only 30 are active in the health system, while the new 6 genetic counselors (3 nurses and 3 psychologists) are expected to finish at the present year (Paneque and Sequeiros 2010).

In Portugal, cancer genetics services are generally integrated in the National Health System (NHS). Regional oncological hospitals and genetics departments of some central hospitals include oncogenetic counseling, as for the case of some public institutes. Some medicine faculties and private entities often assist with clinical and laboratorial expertise. A National Program of Presymptomatic Test and Genetic Counseling was approved and implemented in the mid-nineties, primarily directed for adult neurological onset disorders, and later expanded to other forms of genetic and common diseases (Health General Directory 2004). Since 2005, a specific Portuguese law on genetic information requires that carrier, presymptomatic and susceptibility tests must be preceded by genetic counseling and written informed consent, and any predictive testing should be preceded by a psychosocial evaluation and followed after the delivery of the results.

## 6.2. METHODS

### 6.2.1. Design and procedure

A qualitative study design was used, since the main emphasis was to explore people's experiences and perspetives in an area where little is known to guide research or practice (Glaser and Strauss 1967; McAllister 2011). The research design involved focus groups, a method often used for exploratory purposes such as needs assessment and programmes' evaluation. Its informal group structure intends to encourage participants to speak freely and to engage in discussion aiming to generate ideas and gain insight about a given topic (Piercy and Hertlein 2005). Previous research has indicated that focus groups are suitable to work in the context of clinical genetics services and with genetics professionals (McAllister et al. 2007; McAllister et al. 2010), as well as in a wide range of investigations exploring what service users and carers consider important in providing community services (Unell and Bagshaw 1997). Our initial purpose was to perform focus groups; however, 3 individual interviews were undertaken due to difficulties in finding a common schedule for the focus group. Overall, 3 individual and 6 semistructured focus group interviews were facilitated by the first and third authors (AM, MP), lasting approximately 1 hour. The interview topic guide can be seen in Table 6.1.. One focus group gathered professionals from 2 institutions from the same city. All interviews were digitally recorded with participants consent.

**Table 6.1. Interview guide**

| Topic | Questions |
|---|---|
| Models / principles of practice | *Are you identified with any particular model in your genetic counselling practice? Do you recognize psychosocial support as part of genetic counselling?* |
| Provision of psychosocial support | *How the provision of psychosocial support in this service usually occurs? In which phase of the genetic counselling protocol do you think that family support is more needed?* |
| Needs | *What difficulties do you identify in this service for providing psychosocial support for individuals and families in genetic counselling? What would be necessary to incorporate it here and in genetic counselling protocols in general?* |

### 6.2.2. Recruitment of participants

The sample of genetics health care professionals was recruited from public institutions that delivery cancer genetic counseling in Portugal. A list of public institutions working in cancer genetics were identified using the Health General Directory [13] and by key personnel. An email was sent to each director of the approached genetics healthcare teams inviting the team to participate in an interview regarding the current practice of psychosocial support in oncogenetic counseling. Professionals from different backgrounds were eligible to participate as long as they were part of the multidisciplinary team. Subsequent email contacts were established to arrange and schedule the interviews. Those who agreed to cooperate were given the focus group interview guide containing the topics to be explored for previous knowledge.

### 6.2.3. Participants

10 institutions were contacted; 2 declined to participate. For ethical reasons, motives were not explored. 30 professionals working as part of genetics healthcare teams from 8 institutions (4 major hospitals, 1 regional hospital, 1 oncological hospital and 2 genetics centres) were interviewed. Participants' professional background included: 20 doctors (17 geneticists, from which 4 were interns, 2 oncologists and 1 obstetrician), 3 genetic nurses, 3 psychologists and 4 genetic counselors' trainees (3 psychologists and 1 nurse) (Table 6.2.). Genetics healthcare teams differed between institutions, mainly including geneticists and nurses; in only one case psychosocial professional were integrated in genetics services. One of the genetics centre is mainly devoted to genetic counseling for late onset neurological disorders. Its relevance to be included in this study relies in the fact that it is one of the leading institutions in Portugal to deliver genetic counseling in predictive genetics.

**Table 6.2. Participants' professional background represented in focus groups (*n* = 30)**

| Focus Group | Professional background | Number of participants (*n*= 30) |
|---|---|---|
| 1 | 6 geneticists (1 intern) | 7 |
| | 1 genetic counsellor trainee (psychologist) | |
| 2 | 1 geneticist | 4 |
| | 2 nurses | |
| | 1 genetic counsellor trainee (psychologist) | |
| 3 | 1 obstetrician | 4 |
| | 2 oncologists | |
| | 1 nurse | |
| 4 | 6 geneticists (3 interns) | 7 |
| | 1 genetic counsellor trainee (psychologist) | |
| 5 | 2 psychologists | 2 |
| 6 | 2 geneticists | 3 |
| | 1 genetic counsellor trainee (nurse) | |
| **Individual interview** | | |
| 1 | 1 psychologist | 1 |
| 2 | 1 geneticist | 1 |
| 3 | 1 geneticist | 1 |

### 6.2.4. Data Analysis

Interviews were audio-tapped and fully transcribed and submitted to qualitative content thematic analysis (Strauss and Corbin 1998). Open coding was used to summarize content and representative statements from recurring themes; constant comparison between the emerging themes was carried out by the first author and an independent researcher, to enhance reliability (Strauss and Corbin 1998). Successive coding refinement through repeatedly reviewing the transcripts was made until consensus was reached, aiming to develop content categories; categorization titles were given to similar themes and contents.

## 6.3. RESULTS

Three main themes and their respective subthemes emerged from the content analysis of the interviews transcripts (Table 6.3.).

**Table 6.3. Themes and subthemes derived from the interviews**

| Theme | Subtheme |
|---|---|
| Current practice in cancer genetic counselling | Current practice |
| | Psychosocial delivery |
| | Critical moments for psychosocial adjustment |
| Unmet service needs | Structural needs |
| | Organizational barriers |
| Improving practice | Multidisciplinary teams working within genetics services |
| | Integrated and coordinated care |
| | Training in specific issues |
| | Primary care professionals education in genetics |

### 6.3.1. Current practice in cancer genetic counselling

#### *6.3.1.1. Models of practice*

Participants reported that don't follow any particular theoretical-based approaches, although non-directive counseling was consistently mentioned as a guiding principle in their standards of practice. The main goal is to impartially provide neutral information:

> "*We don't have a model. We provide concise and objetive information in a non-directive fashion; our goal is not what decision the counselee will make, but rather if that decision is made freely and through an informed manner*".

To convey correct and succinct information appeared as the core issue of the genetic counseling provision for the majority of the interviewed professionals'. Great attention is devoted to transmit objetive risk information, suitable of being easily understood by counselees:

> *"We try to assure that all the risk estimates are transmitted in a way that people can understand; we also explore the counselees' motivation and risk perception, and anticipate possible future implications and scenarios, such as prophylactic surgeries when appropriate"*.

Some professionals described an eminently medical-oriented practice, mainly concerned with risk detection and subsequent medical care:

> *"We follow a purely clinical model based on risk measures after collecting the family history and evaluate if the genetic test is pertinent, and then is the counselee who will decide! It is a medical consultation and our role is merely to give correct information and refer surveillance or risk reduction options if appropriate"*.

Other professionals described an experience-based practice. As specific training in genetic counseling skills were not achieved for the majority of genetics healthcare professionals in Portugal, informal networks of colleagues and autodidactic readings were stated as a way to improve these professionals' practice skills, and to debate specific doubts:

> *"We didn't have specific training in genetic counseling (…); we learned while observing the consultations and then through our experience with cosultands (…). We support each other and we try to get updated with books and articles, not only on oncogenetics but also on how to do the counseling and related techniques"*.
>
> *"We don't follow any proper model, we rely in our clinical experience in oncology applying validated risk tables, and we regularly try to share our doubts with our colleagues"*.

Some accounts explicitly point the need to include different approaches in genetics, away from the traditional preventive-curative models:

> *"I think it is important not to adopt the medical-traditionalist model* [in genetic counseling], *which is very paternalistic and therefore not suitable for genetics because we deal with predictive territories"*.

However, some clinicians argued that, culturally, clients' commonly expect directivity from clinicians and some guidance to manage their health problems; so, a transition to a more collaborative role need to be gradually developed.

#### *6.3.1.2. Psychosocial delivery*

All professionals acknowledged psychosocial support as an integrant part of the genetic counseling provision. Genetic counselors' trainees emphasized the importance of performing *a psychosocial-oriented counseling*. However, the majority of professionals reported psychosocial delivery as commonly limited to an informative scope, such as answering to any questions, provision of written leaflets, or drawing illustrative sketches of genetic inheritance patterns:

> *"We always give counselees a resume-letter with detailed information about inherited risk, surveillance or prophylactic options, and relatives potentially at-risk. Besides, counselees can always contact us for doubts"*.
>
> *"We have two kinds of informative sheets for counselees: one to help them understand hereditary cancer risk information, and the other is a decision aid for prophylactic surgery"*.

Also an informative questionnaire to assess social needs (*e.g.*, transportation or financial burden) is administrated by one genetics team, although they think that specific support for these cases is inexistent:

> *"I think that information* [from the questionnaire] *does not help very much because there is nothing more we can do besides our attentive posture (…). So we recently recruited a social worker and now we are trying to envisage a way to include her services"*.

Psychological support was mainly offered for consultands evidencing difficulties while managing their genetic status. Professionals working with late onset neurological disorders follow a pre-symptomatic test protocol, which includes psychological assessment at pre- and post- test counseling. In all institutions, the provision of familial support is rare and when requested occur through individualistic approaches. One genetics team was integrated in a paediatric hospital, where couples were seen for pre-natal and pre-implantation diagnosis or pre-natal screening. This support is assured by psychologists. Their services are usually requested through referral after the genetic counseling consultation, when needed.

> *"People may have psychological support if they ask for it, or we can refer when we identify signs of anxiety or depression; frequently they don't want to, and when they do, they often drop-out".*

Psychosocial support in genetic counseling services is requested through referrals to professionals (psychologists, psychiatrists, or social workers) who are not integrated in the genetics service, but in other departments. Such procedure was stated as prejudicial, because they are already constrained by other demands. Multidisciplinary teams working specifically with cancer patients also helped genetic counseling patients in two institutions:

> *"If needed, we refer for the psychologist or social worker, but it is far for being the ideal because both are mainly dedicated to other departments and specific areas – children with learning disabilities due to a phenylketonuria diagnosis – so often there is a waiting list"*
>
> *AM: And who carry the evaluation of when or if psychosocial support is needed?*
>
> *"We do (…) somehow we try to manage by ourselves the difficult moments in the consultation but it is not enough because there is no psychologist in the service!".*

Some genetics services refer counselees needing psychological support for external services:

> *"The support that we provide is scarce, we have no means. (…) we don't have a psychologist on a permanent basis, so we use to refer for external services, but ideally that should not happen because I believe that some counselees lost themselves in this way; we should offer this service".*

#### *6.3.1.3. Critical moments for psychosocial adjustment*

Three specific moments of the cancer genetic counseling timeline were consistently indicated as commonly posing higher levels of psychosocial distress to counselees and their families: i) waiting for test results, ii) knowing the carrier status, and iii) going for prophylactic surgery.

> *"Waiting for results, results disclosure, and when prophylactic surgeries are suggested, are the moments when counselees often get very anxious and fearful, although some of them don't show it".*

However, professionals' emphasised the whole genetic counseling timeline as potentially stressful:

> *"I think it* [psychosocial support] *should be made available since the very beginning in order to identify possible problems and to prepare the patient for every possible scenario, such as risk reduction options, or even symptoms. All the counseling timeline has potential to disturb counselees and their families"*.

The family developmental phase and the dissemination of genetic risk information among relatives were acknowledged as potentially distressing for families:

> *"People often get distressed when they realise that close relatives may also be at-risk for cancer, especially younger descendants, before 18 or in their twenties"*.
>
> *"We feel sometimes is complicated for people to get in touch with them* [potentially at-risk relatives], *and tell they might be at-risk, whether due to conflicts or distance, or because they fear a bad reaction. Several families fail to continue the genetic study because of this (…); I guess the information that runs in families is in many cases inaccurate"*.

**6.3.2. Unmet service needs**

Professionals described unmet needs that prevent the provision of psychosocial support in their genetic counseling services. A number of current resource issues involving structural needs (lack of funding, confined physical space, and limited qualified human resources) and organizational barriers (lack of a multidisciplinary team working specifically in the genetics service, and little co-ordination between medical specialties involved in cancer genetics care) were identified:

> *"Basically, to do that* [provide psychosocial support in genetics services] *we need funding for recruiting a multidisciplinary team, and physical space. We've made several requests and we are still waiting"*.

One of the clearest messages from genetic counseling teams was the lack of human resources capable of assuring the adequate psychosocial care of patients and families. Professionals stressed the need and the advantages in having a multidisciplinary team integrating psychologists and social workers in genetics services:

> *"I am in this* [genetics] *service on a part-time basis, only 2 days a week. (…) we should have more means to provide psychosocial support, with a psychologist and a social worker"*.
>
> *"We need psychologists and social workers integrated here* [at genetics services] *permanently and not just attending our referrals, because they have to attend referrals from services of all over the hospital. Besides, cancer genetics demand specific needs, tailored to certain moments and decisions"*.

A better coordination among the several medical specialties involved in the scope of cancer genetics was also stressed. Dispersion of services and facilities pose a major limitation for individuals and families' psychosocial accompaniment. This happens due to the inherent nature of cancer genetics and its interdisciplinarity assessments in risk management, where patients may then be under the care of a number of medical specialties as they frequently have to navigate through other hospital departments or even external referrals, often with little coordination between them:

> *"A better articulation between the several medical specialities involved in familial cancer risk within the hospital is essential as complementary services for the genetic counseling consultation; what we use to do is through informal contacts and sometimes it takes too long. When we have to deal with services from outside the hospital it is problematic because the information is often contradictory"*.

Some participants mentioned the need to sensitise policy makers about these barriers for an integrated care in genetics services so these needs may be met:

> *"We need a structure, a service organized under psychosocial principles, with psychologists and genetic counselors, trained professionals who can assure integrated care for patients and their families. We are aware of these limitations but first we need to apply for head hierarchies in order to change policies"*.

### 6.3.3. Improving practice

In the sequence of the mentioned needs, professionals advanced some suggestions to improve the quality of cancer genetics' services. The need to include a multidisciplinary psychosocial care since the initial contacts with the genetics service was outlined as a way to assess current and future difficulties:

> *"A psychosocial consultation prior to genetic counseling should be provided to assess needs and anticipate difficulties; therefore we could track patients and families throughout the genetic counseling timeline"*.

Training for genetics healthcare professionals in particular aspects of genetic counseling were also pointed as beneficial:

> *"Perhaps we could have some training in specific issues, such as how to better communicate with our patients"*.
>
> *"People don't tell us this directly but sometimes we feel consultands have difficulties in talking with relatives, so maybe we could study a functional way to help them to reach them* [family members, potentially at-risk individuals]*"*.

Cancer genetics often demand services of unrelated medical specialties; integration and coordination among medical specialties involved in cancer genetics were stated as a major need.

Participants envisioned ways to keep counselees under ongoing care, even after genetic counseling or testing is complete: a centralized informatics case management services, or a "patient care ombudsmen" were suggested as a way to support patients by overseeing their care:

> *"Oncogenetics demands a great deal of coordination between several medical services, often between different and distant institutions, and we feel that it is difficult to assure that what we recommend is effectively done, even inside the same hospital. Ideally, a sort of patient ombudsmen should exist, to act as an intermediary of the patient care, and a centralized informatics' system to track the patients' path and care agenda (…). These would prevent that patients loose themselves in the NHS"*.

Partnerships with community-based services were also mentioned as means to aid further effectiveness and continuity of care to at-risk patients and families:

> *"Partnerships with community-based services could be a good idea, especially for patients and families from rural and distant areas"*.

The role of primary care professionals was viewed as pivotal for co-ordination purposes, as a great number of oncogenetics consultations result from their referrals. Despite the majority of interviewees use to send a resume-letter about the cancer susceptibility status to the family doctor, through the consultand, education in basic aspects of genetics, including how to perform appropriate referrals, is seen as a relevant aspect for adequate resource management; policy issues were also suggested:

> *"Primary care doctors need training in basic genetics concepts to increase adequate referrals because in many cases those referrals are completely wrong. They need to be sensitized to the Amsterdam criteria and in analyzing the family history, for example"*.
>
> *"Regional health administration institutions should promote training actions or produce and distribute informative material; another idea is to integrate observation in genetic counseling as an internship option for young oncologist interns"*.

Some participants were aware of the recent Master course in genetic counseling in Portugal (some trainees were integrated in the interviewed teams). These professionals observe the future inclusion of trained genetic counselors as a step further for the quality of genetic counseling provision:

> *"I think that the new genetic counselors may play an important role in genetics services in the future. We should adapt our structure in order to incorporate them, but this is a matter for policy maker"*.

## 6.4. DISCUSSION

The themes identified in these focus groups contribute to elucidate the current practice of genetic counseling in the specific field of cancer risk genetics in Portugal. Findings also reflect relevant issues for genetic counseling practice given the growing efforts that have been made to assess standards of practice and education for professionals (Skirton et al. 2010a-b). A model of practice constitutes an important feature for genetic counseling, as it can be used to investigate service provision, to enhance its effectiveness, and can provide a basis for educating adequate skills for professionals (McCarthy Veach et al. 2007).

Our study elicits heterogeneous levels of constraints in the service delivery system and educational needs of professionals, which may provide an exploratory account in identifying education routes for healthcare practitioners working in oncogenetics. This study appears in a particularly important moment in Portugal, since the first generation of Portuguese genetic counselors are completing their master degree training, aiming to train genetic counselors to join multidisciplinary teams at medical genetic services. As some current challenges for the provision of cancer genetic counseling in Portugal are drawn, these professionals' views may contribute for envisioning paths for the integration of genetic counselors in the provision of oncogenetics.

### 6.4.1. Current practice of cancer genetic counselling in Portugal

To assess risk and to consider genetic testing appear as central tasks to the genetic counseling session; later, decision making for prophylactic interventions is considered too, if appropriate. A predominantly informative-based focus guides the genetic counseling agenda; the provision of correct and succinct information about the clinical aspects of the condition and risk estimates objectively and in a neutral manner is the most salient tenet. These results are consistent with some previous studies (Brain et al. 2000; Ellington et al. 2005; Hopwood 2000; Lobb et al. 2003), although very limited if we consider that other psychosocial issues have been received a more broad attention in the scope of oncogenetic research (Andrews et al. 2004; Meiser et al. 2008; Pieterse et al. 2007).

Professionals' stated that they do not follow a "model", although non-directive counseling was mentioned as an important standard of their practice, even if primarily associated to the way how information was provided. In general, professionals' accounts indicate their practice as closely aligned to the teaching/educative model. References to current practice were mainly related to the way information is presented and the confirmation of its comprehension by counselees; other psychosocial-oriented issues were limited. To enhance the comprehensibility of the information, some genetics services provided informative written material; others used risk figures at the consultation. Support materials to the consultation have been described as a useful tool by many

other authors (Lewis et al. 2007). Psychological support is offered if clinicians felt that it would be needed for clients' adaptation, or if the client expressly requests it. This is a serious drawback of current practice if we consider that even clients may not be capable of acknowledging their psychological needs as effectively as they are able to identify their needs for information or education (Davey et al. 2005). Besides, it separates the psychosocial dimension from the counseling process, rather than providing an integrated genetic counseling approach.

At Portuguese centres included in our study, only one of the professional teams included a psychosocial assessment as a routine procedure. Genetics professionals hardly have had formal training in psychosocial issues or counselling skills; psychological support and genetic counseling tend to be perceived as incompatible matters. We believe that in practice, this assumption makes a collaborative approach to genetics services far more difficult. Besides, relevant cultural issues need to be considered here. As the Portuguese population is poorly ranked at educational levels (OECD 2010) and society in general still overweighs' the value of doctors' opinions, expectations of a more horizontal doctor-patient model of relationship are generally subdued in favour of a more traditional one, which may function as a buffer against distress as well.

### 6.4.2. Improving psychosocial support in cancer genetics services: issues affecting the delivery system

Although all professionals were clear about the importance of providing psychosocial support as an integrant component of genetic counseling, difficulties were also pointed on this endeavour.

Structural and organizational needs were the most striking barriers identified on genetics services concerning the delivery of psychosocial support for individuals and their families. Emphasis was drawn on the scarcity of human resources adequately trained to assure quality psychosocial care, particularly psychologists and social workers. As to the present moment, there is no experience of an integrative practice of genetic counselors in Portugal, and valuable core competences on psychosocial support of genetics healthcare professionals are still underestimated. Lack of multidisciplinary teams in genetics services were therefore stressed as a major need, which is aggravated by specific issues as confined physical space or budgetary constraints for personnel hiring. These aspects were also found in a recent study assessing clinical practices among cancer genetic counselors (Wham et al. 2010).

Multidisciplinary teams were rarely stated as available within genetics services; psychosocial professionals, as psychologists and social workers are often dispersed through other hospital services, with limited time for supporting the genetic counseling agenda. Such scenario can result in patients having to attend multiple hospital appointments, which is aggravated if multiple family members are under care. Moreover, it has been stated that the lack of education in interprofessional

collaboration can limit effective team communication and access to appropriate care providers; this can result in gaps, fragmentation and duplication of services (CAPO 2010). Furthermore, coordination of medical specialties involved in cancer genetics is crucial. Individualized oversights of care, centralized informatics' services, and further articulation with community-based facilities are among the suggestions that clearly point the need towards maintaining contact and continuity of care with consultands and their families.

### 6.4.3. Implications for practice and policy

Although participants' assessment of current services stressed several limitations, they did offer concrete examples of what would improve their delivery in oncogenetics. While the importance of an adequately trained multidisciplinary team has been reported, some professionals also mentioned specific needs in the genetic counseling process, such as specific training in communication with families and in how to help them disseminating genetic risk information among relatives. These findings are consistent with previous studies which highlighted the idea that theoretical and practical education is required in order to gain understanding and skills in implementing interventions (Adler and Page 2008; Gotlieb and Wachala 2007; Hack et al. 2005; Jacobsen 2009).

The need to educate primary care physicians and other non-genetics practitioners about genetics has been presented as a priority (CAPO 2010; Skirton et al. 2010b). Lack of training and of confidence of these professionals to carry out medical genetics tasks has been stated (Nippert et al. 2011). Primary care doctors' training in genetics, especially in how to make appropriate referrals, may indeed improve the service, given the amount of patients at only marginally increased risk referred for cancer risk counselling at some of the participant cancer genetics services; genetic testing or even extra surveillance was not indicated for these patients. Furthermore, improving the selectivity of referrals to genetics services can result in reduced time for clients to receive test results, diagnoses, and prophylactic options. Hence, it is relevant to envision what roles non-genetic specialists, and namely primary care clinicians, should assume in the mainstay of cancer genetics healthcare. Besides educational needs, institutional and cultural dynamics need to be taken into account, namely considering the nature of inter- professional and -organizational contacts (Battista et al. 2011).

Concerning the lack of adequately trained psychosocial preofessionals, Portuguese genetic counsellors have recently completed their training and may therefore ease some of the needs professionals' described in the provision of cancer genetics services, namely: i) by assisting with educational and supportive guidance to patients and their families; ii) updating new developments and promoting adequate counseling techniques for members of multidisciplinary teams; and iii) establishing links with other healthcare resources, namely with primary care and community-based

services. This is consistent with a report of a collaborative approach to cancer genetic testing describing an effective use of genetic counselors' expertise in the process of training and supervision of other healthcare practitioners (Cohen et al. 2009). Again, this is not yet the current experience of Portuguese services.

Cancer genetics is a relatively new area of medicine and involves an overlap of genetic and oncological care. There are some common themes and needs in this new area which would be nationally applicable to the provision of oncogenetics services with locally tailored solutions. These are: (i) the need for inter-professional and inter-institutional networks of those involved in oncogenetics care, namely in cancer surveillance, diagnosis, and prophylaxis, from genetics centres to primary care services; and (ii) the urgent need for core competencies and professional guidelines for oncogenetic counseling, and education of medical and allied health professionals in Portugal.

We believe a regulatory document could be helpful at national level integrating oncogenetic specific milestones into psychosocial oncology tenets. By defining what people under cancer risk counseling or diagnosed with cancer and their family members might expect to receive in relation to their psychosocial health care needs, including assessment, evidence based interventions and access to supportive care, the provision of psychosocial support in oncogenetic services might commence to be planned. Accordingly, since needs vary at different points in the cancer genetics *continuum*, people need appropriate regular information regarding psychosocial oncology resources that exist in their communities (Fitch et al. 2008).

Cross-specialist work and models of care involving close interaction between community (primary), secondary and tertiary care have been developed using locally tailored models of working, as is the case of the Canadian Association of Psychosocial Oncology (CAPO 2010) and the English Primary Care Trusts (PCTs), among others (Campbell et al. 2002; Skirton et al. 2010b). In Portugal, policies and standards of practice may begin to be discussed at this point regarding human resources and services organization.

### 6.4.4. Future research directions

This study presented an exploratory account on the current practice of cancer genetic counseling in a relevant sample of Portuguese oncogenetic services. Future research is needed to understand current models of practice and their implication on quality issues of Portuguese oncogenetic services. In order to identify organizational principles for cancer genetics services and to produce standards of practice for Portuguese genetics healthcare professionals, more focus groups should be conducted involving all the public services that deliver cancer genetic counseling (or supported by the medical professional society). Also, other stakeholders' views, such as service users, patient representatives, and health authorities need to be acknowledged in this effort in order to prepare

changes towards a more effective care. It would also be of interest to investigate how the inclusion of genetic counselors in the mainstay of genetics services is envisioned by the key stakeholders, and how they would improve current practice in a short-term period.

## 6.5. CONCLUSION

This study enfolds an opportunity to begin a wider discussion on some of the challenges that cancer genetic counseling face in Portugal. Findings may contribute to raise awareness on the practice of oncogenetic counseling and on the roles of cancer genetic services in the context of post-genomic healthcare.

We examined the ways in which various genetics healthcare professionals described their models of practice and provision of psychosocial support in the scope of cancer genetic counseling, as well as resource issues affecting the delivery system. Dominant constraints need to be acknowledged in order to improve quality of practice at oncogenetic services. Lack of trained workforce and coordination of care appear to be problematic, preventing effective psychosocial support for counselees and their families, and cost-effective care. Besides, limited funding permeates these barriers; scarce time and rigidified institutional structures limit the role of non-genetics clinicians. Beyond training and material constraints are issues of culture.

Cancer genetics encapsulates both genetic and oncology expertise, representing distinctive professional communities and institutionalized roles, as well as diverse interactions between psychosocial and biomedical healthcare and the genetic technologies (Miller et al. 2008). Cancer genetics healthcare in Portugal certainly needs solutions involving instrumental dimensions and, perhaps more importantly, also needs an organization of cancer genetics services and adequate professionals' training, including a reconfiguration of professional roles, in order to integrate collaborative standards of care and adequate forms of access for patients.

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# CONCLUSÕES GERAIS

## PARA UMA ORIENTAÇÃO FAMILIAR NO ACONSELHAMENTO GENÉTICO: EXPERIÊNCIA, INTERVENÇÃO E CONTEXTO

Globalmente, esta investigação procurou integrar os aspetos experienciais presentes nos subsistemas da unidade de avaliação que Góngora (2004) propôs para a compreensão sistémica de doenças graves, aqui transposta para o espectro do risco genético: interação interdependente entre indivíduo, família / rede social e serviços de saúde, inscrita num tecido sociocultural que influencia os significados e ações em relação à doença e aos cuidados de saúde.

Nos últimos anos multiplicaram-se as perspetivas de pesquisa no âmbito da genética psicossocial, que sublinham a necessidade de conhecer o impacto individual e familiar decorrente da nova genética (nomeadamente, associados ao risco, aconselhamento e testes genéticos), com o intuito de melhorar os serviços clínicos disponíveis. No editorial de um número especial da revista *Familial Cancer*, dedicado aos desafios emergentes na avaliação do risco e aconselhamento oncogenético, Eeles, Purland, Maher e Evans (2007) enfatizaram a necessidade de incorporar uma dimensão biopsicossocial na provisão dos cuidados de saúde através da integração de uma rede multidisciplinar de profissionais com formação adequada, colaborativamente atuantes nos serviços de genética e nos serviços de saúde primários e comunidade. Os estudos nesta tese procuram integrar os desafios mencionados através de pesquisas interligadas que abrangem as respostas de cada subsistema no confronto com as exigências da suscetibilidade oncogenética.

A configuração da tese engloba capítulos compostos por estudos que apresentam conclusões específicas relativamente aos resultados e à sua discussão, bem como reflexões sobre as limitações decorrentes da metodologia. As conclusões gerais, que a seguir se apresentam, procuram conjugar

os principais resultados desses estudos e facultar uma pespectiva reenquadradora das suas conclusões, culminando com recomendações para a intervenção psicossocial no âmbito do aconselhamento oncogenético. Providenciam-se, igualmente, os principais contributos, limitações e perspetivas de pesquisa futura, bem como uma reflexão crítica do desenho metodológico adotado ao longo da investigação.

## 1. SÍNTESE INTEGRADORA DOS PRINCIPAIS RESULTADOS E CONCLUSÕES

O risco e aconselhamento oncogenético são uma experiência intergeracional que inclui trajetórias relacionais que extravasam as ligações biogenéticas. Trata-se de um procedimento médico útil e necessário na promoção de maior controlabilidade sobre a gestão da saúde individual e de outros familiares e norteado por valores altruístas. A informação genética é percebida pelas famílias como "complexa" e "científica". A experiência individual e familiar indica dificuldades e necessidades específicas, que intersetam o desenvolvimento e identidade individual e familiar e são permeadas por um complexo emocional que condiciona o ajustamento psicológico individual e o planeamento da vida familiar. Foram identificados os seguintes momentos críticos do processo de aconselhamento oncogenético: hiato temporal entre a realização do teste genético e o conhecimento dos resultados; confirmação do estatuto de portador de mutações genéticas deletérias; tomada de decisão quanto a intervenções profiláticas de redução do risco; e transmissão da informação genética aos familiares.

A compreensão individual da informação genética e a sua disseminação intrafamiliar envolve dificuldades evidentes que carecem de formas de apoio. A complexidade inerente à informação genética e às suas repercussões individuais e familiares requer serviços de apoio psicossocial adequados às necessidades que suscita, adaptadas à população e disponibilizadas ao longo da linha temporal do aconselhamento genético. A incorporação de uma orientação familiar nos serviços de aconselhamento genético afigura-se necessária. A conceptualização da experiêcnia (*cf*. Fig. 7.1., pág. 206) pode constituir-se como modelo prospetivo e referência compreensiva ao planeamento e prestação de cuidados de saúde dirigidos a indivíduos e famílias envolvidos neste processo.

Os dados suportam a adoção de modelos de aconselhamento genético que enfatizam a dimensão psicoeducativa; ou seja, um modelo de prática centrado na facilitação do uma relação terapêutica que potencie o uso personalizado da informação genética e a autodeterminação, minimizando o *stress*. Adicionalmente, as intervenções multifamiliares revelam um potencial complementar ao aconselhamento genético; os dados destacam o seu efeito facilitador no bem-estar psicossocial e empoderamento dos participantes, sobretudo através do reforço de redes de apoio mútuo,

expressividade emocional e sentimento de controlo sobre a gestão da saúde. As considerações metodológicas decorrentes da implementação das intervenções multifamiliares sugerem a necessidade de flexibilizar o seu desenho estrutural e, numa perspetiva mais alargada, diversificar as modalidades de apoio psicossocial a indivíduos e famílias de acordo com as necessidades identificadas (multifamiliares, unifamiliares e individuais).

O aconselhamento oncogenético segue premissas biomédicas e privilegia um modelo educativo ou de ensino, predominantemente centrado na disponibilização de informação. O enfoque na família é geralmente inexistente. Os serviços de genética revelaram carências estruturais e organizacionais que obstam à disponibilização de intervenções psicossociais centradas na família, nomeadamente, a ausência de equipas multidisciplinares. Tais constrangimentos decorrem de limitações macro-contextuais, incluindo escassez de financiamento das instituições públicas e inexistência de um plano integrado de cuidados em oncogenética. Uma orientação psicossocial nos cuidados de saúde requer a redefinição do contexto organizacional e intersectorial dos serviços no sentido da formação adequada dos profissionais (incluindo um plano de formação básica aos profissionais de saúde primária em temas relativos à genética clínica e ao aconselhamento genético), da integração colaborativa de recursos humanos multidisciplinares, e da agilização do envolvimento dos cuidados primários e recursos comunitários na rede de serviços de apoio psicossocial.

## 2. ROTEIRO METODOLÓGICO: CONSIDERAÇÕES

O alargamento do enfoque do estudo que norteou os trabalhos da investigação (da pesquisa da experiência individual à da família, até à exploração das respostas do sistema de saúde através dos profissionais) refletiu a hierarquização ecossistémica que postula a incorporação de unidades de análise cada vez mais alargadas e complexas nos processos de significação (Bateson, 2000). A evolução da investigação aspirou a um conhecimento multidimensional, capaz de captar as articulações entre partes que compõem o todo ecossistémico em relação. Porém, como adverte Morin (2003), complexidade não é sinónimo de omnisciência, pelo que paralelamente à ambição de abarcar a complexidade e um conhecimento não-parcelar e não-redutor deverá reconhecer-se o seu caráter incompleto e inacabado.

Podem ser gizadas analogias entre as considerações epistemológicas anteriores e a investigação científica aplicada ao cancro. Com a emergência da nova genética, a investigação oncológica constitui-se como um objeto de *fronteira*, reapropriando diferentes contextos, agentes sociais e instituições. Em suma, representa o que Nunes (1996) designa por escala heterogénea, pois evoluiu

da "molecularização"[32], isto é, de uma questão específica da medicina e da saúde, até às suas vastas implicações morais e sociais. Das interações entre a biologia molecular e genética e destas com o tecido sociocultural, ocorreu o alargamento do elenco de "porta-vozes" do problema, permitindo deste modo "*reafirmar as continuidades entre a ciência, os seus objetos e protagonistas, (…) e os espaços e contextos extracientíficos*" (Nunes, 1996: 42).

Os estudos incluídos nesta tese adotam metodologias qualitativas de recolha (*focus group*, entrevistas individuais e familiares) e análise dos dados (*grounded theory*). O desenho metodológico seguiu a premissa participativa que preconiza a auscultação dos utilizadores como aspeto central no planeamento da provisão de serviços de saúde (Brydon-Miller, 2004; Gustavsen, 2001). A escolha do desenho metodológico num processo de investigação envolve a ponderação de potenciais benefícios e limitações, bem como um questionamento prévio sobre a exequibilidade dos objetivos, considerando fatores institucionais, temporais e financeiros. Mas envolve também a auscultação das ressonâncias pessoais do investigador face ao objeto de estudo que, em grande medida, conjuram o caminho metodológico a percorrer. Delinear um roteiro de pesquisa integrado e coerente constituiu, por conseguinte, um dos maiores desafios desta investigação, ancorado no desejo de contribuir através de formas de articulação entre pesquisa e intervenção facilitadoras de um diálogo participativo com os vários atores no domínio da saúde.

A complexidade dos desafios que indivíduos e famílias enfrentam nos cuidados de saúde na era genómica, associado a um enfoque sistémico na sua conceptualização na gestão da vida, impeliu o investigador (autor desta tese) a "mergulhar" na realidade em apreço que encapsulava o objeto de estudo: o espaço clínico onde a prática do aconselhamento genético em cancros hereditários teria lugar; o contacto com os profissionais; a consulta e o seu contexto administrativo; enfim, as dinâmicas pragmáticas e tácitas com que indivíduos e famílias se confrontam no contexto do aconselhamento oncogenético. Durante cerca de nove meses, por três horas semanais, o investigador presenciou as sessões de aconselhamento genético enquanto observador, estendendo esta atividade para além dos requisitos metodológicos relacionados com o recrutamento de participantes do primeiro estudo desta tese. Esta experiência proporcionou a imersão nas múltiplas realidades do contexto fenomenológico do objeto de estudo, contemplando indivíduos e familiares que os acompanhavam, profissionais, espaço clínico e institucional e o próprio investigador. O confronto desta realidade com um "olhar" científico, externo e ético (complementar à perspetiva

[32] Nunes (1996) afirma que a "molecularização" do cancro designa a "fixação" do seu móbil de investigação à determinação genética decorrente da investigação académica e da indústria biotecnológica, e de um progressivo abandono da investigação e prevenção do cancro como problema associado às agressões ambientais. Como consequência da associação à "escala molecular", o autor alude à "desterritorialização", isto é, à passagem da atenção científica à sua organo-especificidade para o âmbito mais geral da biologia molecular e da genómica, como atesta o Projeto do Genoma Humano.

émica[33]), potenciou o reconhecimento de padrões e a aproximação à sua compreensão conceptual, desvelando as premissas que nortearam esta investigação e seu desenho metodológico.

A natureza exploratória do estudo, incidindo num tema ainda pouco pesquisado, cujas variáveis carecem de adequada identificação e clarificação, aliada a integrar uma amostra relativamente reduzida de participantes, não significativa e eventualmente não generalizável, fez-nos optar por metodologias qualitativas desde o início da investigação. Consideramos estas metodologias como um veículo privilegiado para captar a complexidade inerente a uma abordagem intersistémica do aconselhamento genético (Eunupu, 1997; McDaniel, 2005; Rolland & Williams, 2005). Seguindo um corpo de investigação bem estabelecido em genética psicossocial (Beeson, 1997; Grubs & Piantanida, 2010; McAllister, 2001; Michie, McDonald, & Marteau, 1996; Peters, McAllister, & Rubinstein, 2001), a utilização de métodos da *grounded theory* (Glaser & Strauss, 1967) foi transversal aos estudos. Esta metodologia adequa-se aos pressupostos e objetivos da investigação: mais do que identificar reações ou aquilatar a extensão de impactos psicológicos ou sintomatológicos, os objetivos centram-se na caracterização das perceções e da experiência de grupos específicos envolvidos numa área substantiva de pesquisa (indivíduos e famílias participantes no aconselhamento oncogenético, participantes num programa de intervenção e profissionais de saúde envolvidos na provisão de aconselhamento oncogenético).

A *grounded theory* permite uma conceptualização teórica fundamentada nos dados (Glaser & Strauss, 1967). Trata-se de um método indutivo de identificação de padrões recorrentes de relações entre processos, que enfatiza o conhecimento do mundo fenomenológico e social dos participantes. Esta abordagem desenvolve uma descrição conceptual baseada na evidência empírica dos dados e não na construção *a priori* de um quadro de referência que implica a seleção de variáveis (Strauss & Corbin, 1998). O seu potencial valor explicativo e preditivo advém de evidenciar o contexto experiencial dos intervenientes perante um conjunto de circunstâncias, possibilitando o desenvolvimento de intervenções que respondam às necessidades patenteadas (Grubs & Piantanida, 2010). Contudo, como advertem Beeson (1997) e McAllister (2001), há a considerar que a matriz indutiva e interpretativa da *grounded theory* remete os dados para territórios dificilmente objetiváveis. Assim, a análise depende largamente da constelação de capacidades do investigador para gerar conceitos significativos a partir da codificação e categorização temática do material discursivo. Acrescem, ainda, dificuldades em responder a tipos de questões comparativas específicas e em obter um julgamento independente dos resultados. Estes fatores situam-se nos antípodas das assunções positivistas herdeiras da tradição hipotético-dedutiva que conceptualiza a

[33] A perspetiva émica, em pesquisa etnográfica, designa o que Peters, McAllister, e Rubinstein (2001: 136), num estudo desenvolvido num hospital oncológico dos E.U.A., descrevem como: "*the insider view of a groups reality. (…) Whereas the emic perspetive most closely approaches the phenomenological world of the group being studied, the etic perspetive refers to the external, scientific perspetive. The emic view helps the researcher understand why people think and behave as they do*".

investigação científica a partir de premissas de objetividade, causalidade, validade e generabilidade, que consubstanciam o conhecimento científico como observável, mensurável e quantificável (Patton, 1990). Neste ponto, é relevante realçar que as assunções decorrentes do quadro compreensivo facultado por esta investigação possuem um valor heurístico[34], isto é, não traduzem a *realidade*[35], mas mecanismos conceptuais de interpretação de um fenómeno; "*heuristics are concerned with meanings, not measurements; with essence, not appearance; with quality, not quantity; with experience, not behavior*" (Douglas & Moustakas, 1984: 42, *in* Patton, 1990: 71).

Barney Glaser e Anselm Strauss, os fundadores da *grounded theory*, recomendam que qualquer teoria fundamentada nos dados (ou modelo conceptual) seja confrontada e "avaliada" quanto à sua validade e utilidade por peritos na área de investigação em apreço (Glaser & Strauss, 1967). A arbitragem científica a que os estudos apresentados foram sujeitos decorrente da submissão para publicação em revistas científicas, bem como o escrutínio formal e informal inerente à apresentação dos dados em encontros científicos[36], norteou a prossecução desse desígnio.

## 3. PRINCIPAIS CONTRIBUTOS DA INVESTIGAÇÃO

A principal finalidade desta tese envolve aprofundar o conhecimento sobre a experiência do risco e aconselhamento oncogenético a nível individual e familiar, associando-o ao desenvolvimento de uma intervenção de apoio psicossocial e à organização dos cuidados de saúde na era (pós)genómica.

Independentemente das vantagens e desvantagens associadas ao desenho metodológico escolhido e ao mérito do enfoque da pesquisa, o principal contributo desta investigação consiste na tentativa de: conceptualizar e compreender a experiência individual e familiar do aconselhamento oncogenético (*cf*. capítulo I); facultar dados para o desenvolvimento, implementação e avaliação de um programa psicoeducativo multifamiliar, enquanto instrumento de apoio psicossocial a

[34] Grubbs e Piantanida (2010: 102) descrevem as heurísticas como *" (…) conceptual devices (*e.g.*, principles, guidelines, typologies, models, hypotheses) depicting complex phenomena. As such, heuristics serve to further discourse and inquiry by offering coherent portrayals of phenomena. Heuristics are not claimed as verifiable accounts that correspond directly to an external, objetive reality"*.

[35] A epistemologia sistémica da cibernética de segunda ordem, designadamente a que deriva do construcionismo social, indica que os conceitos de "verdade", "objetividade" e "realidade" relacionam-se com uma construção social e consensual que resultam viáveis para os seus "construtores", isto é, a realidade não é única mas *multiverso*; ou seja, é uma coconstrução que resulta da interação recursiva permanente entre os intervenientes intra- e extrassistémicos, incluindo o observador e as suas idiossincrasias (Almeida Costa, 1994). Esta perspetiva difere da visão clássica que postulava uma "única" realidade objetiva envolvendo a descrição completa do sistema *tal como ele é*, independentemente da decisão sobre *como* esse sistema é observado (Prigogine & Stengers, 1985).

[36] Alguns estudos desta investigação (ou versões parciais dos mesmos) foram apresentados em encontros científicos: Mendes, Chiquelho, Santos, & Sousa (2010); Mendes, Chiquelho, Santos, & Sousa (2011); Mendes, Guerra, Rodriques, & Sousa (2009), Mendes, Patrão, Guerra, & Sousa (2008) e Mendes & Sousa (2011).

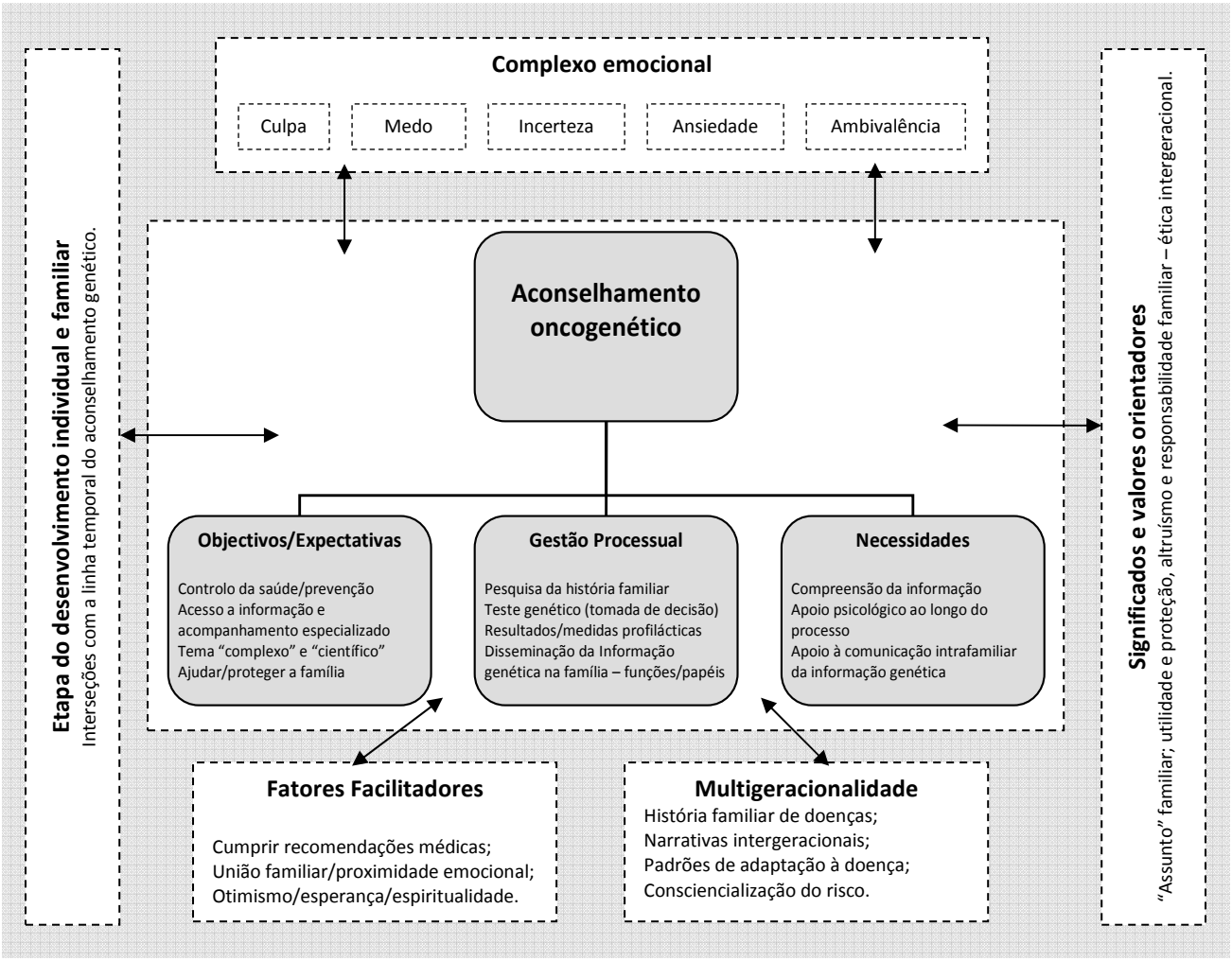
indivíduos e seus familiares com suscetibilidade acrescida a cancros hereditários (*cf*. capítulo II); e integrar a perspetiva dos profissionais em relação ao exemplo de intervenção implementado e à prática do aconselhamento oncogenético, em particular na sua dimensão psicossocial (*cf*. capítulo III). Ou seja, esta investigação contribui para o conhecimento dos processos experienciais inerentes ao aconselhamento oncogenético, relevante para o desenvolvimento da vertente compreensiva do contexto psicossocial que lhe está subjacente, nomeadamente através da depuração de modelos teórico-conceptuais e de intervenções clínicas destinadas a potenciar o bem-estar dos indivíduos e famílias envolvidos. Os dados providenciam, ainda, referências sobre a provisão de aconselhamento oncogenético a nível nacional, num momento particularmente importante no contexto europeu, em que têm sido envidados esforços para a harmonização das práticas de e delineados modelos de educação dos profissionais, aspetos que se relacionam com a crescente necessidade de regulamentação da profissão (Skirton, Lewis, Kent, Coviello, & the members of Eurogentest Unit 6 and ESHG Education Committee, 2010).

### 3.1. Múltiplas dimensões da experiência

Os estudos integrados no capítulo I incluíram a perspetiva de indivíduos e famílias para aceder à compreensão da adaptação psicossocial ao aconselhamento oncogenético. A dimensão experiencial obtida permitiu clarificar padrões de respostas, processos emocionais e relacionais, necessidades e significados associados às várias dimensões da experiência individual e familiar do aconselhamento oncogenético e da gestão do risco. Os resultados contribuem para a literatura, pois disponibilizam a perspetiva familiar, e não somente individual, sobre o aconselhamento genético.

Dada a tendência crescente para a integração do diagnóstico genético de doenças comuns nos cuidados de saúde primários (como os cancros, *cf*. Battista, Blancquaert, Laberge, van Schendel, & Leduc, 2011), é crível que, cada vez mais, as famílias cuja história familiar seja pautada por casos de doença oncológica em várias gerações se deparem com o aconselhamento oncogenético. O quadro experiencial apresentado pode providenciar um referencial normativo das exigências instrumentais, processuais e emocionais de um procedimento médico face ao qual as famílias tenderão a confrontar-se.

**Fig. 7.1. Aconselhamento oncogenético: concetualização da experiência**




### *3.1.1. Quando (o)corre na família: adaptação familiar ao risco oncogenético*

A dimensão hereditária e o efeito cumulativo do historial de doenças na família moldam expectativas e padrões de adaptação à doença e ao risco, contribuindo para que a consciencialização do risco no sistema familiar adquira uma trajetória intergeracional. A ameaça do cancro assume um padrão familiar, que liga passado, presente e futuro e se repercute na identidade familiar. A perceção do risco envolve interpretações construídas a partir do legado narrativo intergeracional, fundadas em crenças de explicação da hereditariedade que descrevem processos atribucionais de criação de significado para a vulnerabilidade face a um futuro temido e ameaçador. O aconselhamento oncogenético é motivado pela prevenção da saúde individual e familiar; aspetos individuais e interpessoais estão na base desse envolvimento, designadamente a obtenção de maior controlo sobre a gestão da saúde e contribuir para a protecção de outros familiares potencialmente em risco. A formulação de objetivos traduz a expectativa em obter acompanhamento médico para aceder a informação relevante sobre um tema "complexo" e "científico", que permita atuar na

prevenção da saúde do próprio e dos familiares identificados como potencialmente em risco (em especial os descendentes). O encorajamento por parte de familiares, em particular dos elementos das gerações mais idosas, desempenha um papel relevante.

A gestão psicossocial dos pontos nodais do aconselhamento oncogenético assume-se como um processo relacional. Após a pesquisa da história médica familiar, a experiência caracteriza-se pela tomada de decisões quanto à realização do teste genético (e consequente conhecimento do estatuto genético decorrente dos resultados) e para intervenções profiláticas de redução de risco, bem como a transmissão / disseminação da informação genética aos familiares potencialmente em risco. Com efeito, foram identificadas algumas regularidades no modo como os sistemas familiares organizam as suas respostas face à gestão processual da informação genética, tendo emergido as seguintes funções: "guardião da informação genética", "pesquisador", "disseminador" e "bloqueador". Os elementos da geração mais idosa foram descritos como tendendo a assumir o papel de "guardiães da informação genética" (e da a saúde em geral), sendo procurados aquando da recolha de informação sobre os cancros na família para a construção do *pedigree*. O "pesquisador" tende a acumular as suas funções com as de "disseminador da informação genética", sendo este duplo papel geralmente assegurado por indivíduos que já tiveram cancro ou pelo o probando (i.e., o indivíduo através do qual se inicia o estudo genético da família). As mulheres destacam-se enquanto mediadoras e facilitadoras do contacto intrafamiliar; o seu papel na manutenção dos laços entre familiares, cuja relação é pautada pela distância (física ou emocional) ou conflito, destaca igualmente a sua função de fonte de apoio emocional. Os "bloqueadores", apesar de identificados, estiveram ausentes das entrevistas familiares, pelo que se infere a existência de uma relação entre a assunção deste papel e o maior afastamento relacional com os demais elementos da família, aspecto eventualmente também relacionado com um menor acesso à informação genética.

O confronto com a informação genética e os procedimentos médicos associados não são uma experiência emocionalmente neutra. Um complexo emocional heterogéneo permeia as várias fases do processo de aconselhamento e risco oncogenético e reverbera de modo diferenciado nos vários elementos da família. Culpa, medo e ambivalência configuram um sentimento geral de vulnerabilidade que acompanha reações de depressão e ansiedade perante os vários pontos nodais do aconselhamento genético. Alguns momentos exacerbam o complexo emocional: hiato temporal que medeia a realização do teste genético e o conhecimento do resultado; confirmação do estatuto de portador de mutaç(ões)ão genética(s); tomada de decisão face à realização de cirurgias profiláticas de redução de risco.

Existem necessidades de apoio psicossocial que se estendem, de modo variável no foco e na intensidade, ao longo do processo de aconselhamento genético, inclusive durante o período pós-teste. Desde o início, o acesso a informação médica é determinante na tomada de decisões

informadas e para a progressiva integração dos possíveis cenários pós-teste, bem como para maior capacitação na transmissão e disseminação de informação genética relevante aos familiares potencialmente em risco. Neste ponto, o maior desafio reside na compreensão da informação veiculada e estimativas de risco. A comunicação com familiares foi mencionada como uma dificuldade específica, pois alguns fatores (como distância geográfica ou emocional, existência de conflitos, grau de compreensão da informação genética e capacidade de a explicar, a par da capacidade e interesse em recebê-la) obstam à eficácia deste processo.

Indivíduos e famílias identificaram fatores facilitadores da adaptação ao aconselhamento oncogenético: união familiar através da expressividade emocional, otimismo e adesão às recomendações médicas facilitam a adaptação psicossocial. Estes fatores parecem atuar num plano heurístico que alivia a carga emocional inerente à incerteza do risco oncogenético, ao mesmo tempo que fortalece os laços relacionais. Seguir as recomendações médicas proporciona segurança, sendo percebido como um comportamento modelar para a restante família, em especial para as gerações mais novas. A ênfase na espiritualidade contribui para o incremento de controlabilidade perante a incerteza subjacente à informação genética.

A experiência é enquadrada num *continuum* espacio-temporal que interseta os vários momentos e pontos nodais que definem o processo de aconselhamento oncogenético e a fase de desenvolvimento individual e familiar. Tais justaposições, sobretudo se coincidentes com períodos de transição evolutiva do ciclo vital, são suscetíveis de desencadear bloqueios desenvolvimentais e amplificar dificuldades.

Indivíduos e famílias atribuem significados de utilidade e proteção ao aconselhamento oncogenético. Uma perspetiva altruísta norteia a experiência pela possibilidade de contribuir para a saúde da família e de cuidar das gerações mais novas, garantindo uma equiparação de oportunidades quanto ao acompanhamento médico. Valores morais-relacionais orientam a partilha da informação com outros familiares sobre os potenciais riscos genéticos e o envolvimento em medidas preventivas e profiláticas de redução do risco. Emerge uma ética intergeracional que molda o envolvimento familiar, percebido como uma responsabilidade individual e um dever moral para com a família. Trata-se de um "assunto familiar", particularmente sentido na família nuclear e em relação a familiares com ligações biológicas em primeiro grau, embora extensível à família alargada.

### *3.1.2. Percursos da informação genética: identificação de funções e papéis familiares*

A exploração das funções envolvidas no processo de transmissão da informação genética não figura nos objetivos específicos desta investigação, mas os resultados obtidos são consistentes com

a literatura quanto a: implicação nos processos comunicacionais intrafamiliares (Ashida *et al*., 2011; Koehly *et al*., 2009; Peters *et al*., 2011); e fatores emocionais, relacionais e de género revelados (Nycum, Avard, & Knoppers, 2009). O estudo das redes sociais da família multigeracional e das relações intergeracionais revela igualmente funções e papéis familiares com características equiparáveis, nomeadamente o "guardião das memórias familiares" e o "elo de ligação" (Vicente, 2010), corroborando a natureza multigeracional da experiência de aconselhamento oncogenético, que envolve a família nuclear corresidente, família de origem e família alargada.

A relevância da comunicação intrafamiliar no contexto do estudo da família e aconselhamento genético advém de indivíduos potencialmente em risco para desenvolverem uma patologia hereditária poderem aceder a informação sobre a sua potencial condição de acrescida predisposição à doença, permitindo que equacionem decisões *informadas* de gestão individual da saúde. Os dados relativos às funções e papéis familiares sugerem que a inclusão de elementos pertencentes à família de origem e às gerações mais idosas pode ter contributo decisivo na eficácia da transmissão de informação genética, bem como na sensibilização à adoção de medidas preventivas de vigilância e comportamentos mais saudáveis (Ashida *et al*., 2011; Ersig *et al*., 2009; McCaan *et al*., 2009). A sua influência simbólica e potencial papel aglutinador dotam os mais velhos de centralidade na gestão familiar de crises, como perante o espectro do risco hereditário. As pessoas idosas da família são frequentemente as primeiras a ser consultadas quando surge uma doença, sendo cruciais na gestão das expectativas e tomada de decisões face a tratamentos e na prestação de suporte emocional (Forrest *et al*., 2003).

### 3.2. Programa de intervenção psicoeducativa multifamiliar

Os estudos do capítulo II desta tese centram-se na implementação e avaliação de um programa de intervenção dirigido a famílias com risco oncogenético acrescido, desenhado a partir de levantamento de necessidades prévio e de dados dos estudos anteriores. Adotou-se uma abordagem psicoeducativa num formato multifamiliar, um modelo de intervenção estabelecido no âmbito das intervenções familiares em contexto grupal (Fadden, 1998; Gonzalez & Steinglass, 2002). A literatura é profícua a destacar a adequação de grupos de discussão multifamílias na adaptação ao risco genético (Esplen, 2011; McDaniel, Rolland, Feetham, & Miller, 2006; Werner-Lin, 2007). A intervenção foi avaliada qualitativamente pelos participantes e por profissionais do aconselhamento genético.

Os programas evidenciaram uma estrutura e conteúdos geralmente adequados às necessidades dos participantes. O enfoque da intervenção, isto é, a facilitação de recursos de apoio psicossocial e o formato multifamiliar, foram mencionados como recursos importantes para o bem-estar dos

participantes. A partilha de experiências entre famílias a viverem circunstâncias e desafios semelhantes e a possibilidade de acesso a informação médica de forma mais pormenorizada e num contexto de maior informalidade, sobressaem como contributos da intervenção. O suporte mútuo e a facilitação da expressividade emocional, sentimentos de empatia e compreensão das dificuldades proporcionaram, simultaneamente, a normalização dos sentimentos e a diminuição do isolamento social e da estigmatização. A repercussão positiva da participação no programa veiculada pelos participantes consolida os grupos de apoio como relevantes na mitigação dos efeitos psicossociais da suscetibilidade acrescida a doenças hereditárias. Participantes em grupos de apoio para indivíduos em risco para a Doença de Huntington reportaram níveis elevados de *stress* e desconforto perante o contexto grupal, designadamente, a possível antevisão das dificuldades no seu futuro (Lowit & Van Teijlingen, 2005), embora neste caso as desvantagens apontadas possam ser atribuíveis ao prognóstico da doença e à ausência de tratamentos disponíveis.

Os benefícios reportados estão geralmente de acordo com a literatura, que aponta os grupos de apoio como uma oportunidade para incrementar recursos familiares (*coping*) no confronto com a doença, em particular em contextos de menor abertura à comunicação intrafamiliar (Plumridge, Metcalfe, Coad, & Gill, 2011). Os resultados indicam ainda que a intervenção implementada pode constituir um contributo na capacitação psicossocial de indivíduos e famílias face ao confronto com a suscetibilidade genética. O seu impacto ao nível do bem-estar emocional, acesso a informação médica e decisões sobre a gestão da saúde, corroboram a perspetiva de empoderamento (*empowerment*). Não obstante a controvérsia em torno da delimitação conceptual do conceito (Ajoulat, d'Hoore, & Decache, 2007), o empoderamento figura como um recente indicador da qualidade da provisão dos serviços de genética e cuja relevância tem vindo a ser destacada na adaptação ao risco genético (McAllister, 2010). A sua depuração conceptual tem sido encetada e os seus efeitos traduzem-se no sentido da autodeterminação dos consultandos e suas famílias, sobretudo através do reforço das suas competências psicossociais para lidarem com uma situação adversa como o risco genético. As suas dimensões incluem a regulação emocional, controlo cognitivo (conhecimento e compreensão da situação), controlo decisório (apoio à tomada de decisão), controlo comportamental (instrumentalidade) e esperança / orientação futura (McAllister, 2010).

Os dados sugerem que os grupos de discussão multifamílias são potenciadores de canais comunicacionais dotados de maior fluidez e abertura. As famílias têm oportunidade de ativar narrativas e significados para a vivência do risco de modo mais flexível e multidimensional (Werner-Lin & Gardner, 2009). O incremento de novas de ligações extrafamiliares sugere a importância do programa enquanto veículo promotor de novos contactos sociais. A nível familiar, a união e a maior proximidade emocional traduzem movimentos estruturais centrípetos,

característicos de sistemas familiares em contextos de crise ou forte ambiguidade. Trata-se de um movimento de preservação da vida do sistema que visa responder à necessidade de reestruturação de papéis e funções familiares (Rolland, 1994). As condições crónicas de doença, como as hereditárias, tendem a comprometer algumas características estruturais e atributos do vínculo relacional da rede social pessoal. Assim, algumas funções específicas de suporte social, como o apoio informativo, o acesso a novos contactos e a possibilidade de encetar interações recíprocas, são relevantes no bem-estar psicossocial do indivíduo (Alarcão & Sousa, 2007; Guadalupe, 2009). O acesso a recursos informativos foi providenciado por duas vias: informação médica disponibilizada pelo profissional de aconselhamento genético, incluindo a discussão de dúvidas em *fórum* alargado; e imersão nas experiências partilhadas por outras famílias. Neste aspeto, foi possível observar a clarificação da tomada de decisão quanto à realização de tratamentos profiláticos, apontando para maiores índices de confiança e conforto na gestão da saúde.

As intervenções psicoeducativas estão há muito difundidas no âmbito do risco oncogenético, mas a literatura é escassa na descrição de intervenções multifamiliares, ou seja, que promovem o envolvimento de outros elementos da família para além do consultando; em suma, capazes de abarcar o contexto holístico da sua interface médica, individual e familiar. A sua pertinência advém do renovado interesse nos cuidados de saúde centrados na família (Steinglass, 2006) e, no caso da nova genética e da medicina preditiva, do caráter familiar e eminentemente relacional das doenças genéticas e do papel proactivo atribuído a indivíduos e suas famílias na monitorização da saúde.

### 3.3. Desafios à incorporação de uma orientação centrada na família nos serviços de genética

Esta investigação procura contribuir para o desenvolvimento de uma abordagem psicossocial nos protocolos do aconselhamento genético, como forma de apoio complementar ao seu enfoque biomédico e tendencialmente individualizado (Capelli *et al*., 2009). Através da auscultação da perspetiva dos profissionais que atuam na esfera do aconselhamento genético, foi possível estabelecer uma triangulação de dados sobre a avaliação da intervenção multifamiliar aplicada e, paralelamente, elencar necessidades para a implementação de uma orientação centrada na família nos serviços de genética.

Um dos contributos dos estudos do capítulo III foi a possibilidade de envolver os profissionais num esforço reflexivo sobre a sua prática num momento particularmente relevante para o aconselhamento genético a nível europeu, em que modelos de prestação de serviços têm sido discutidos (Battista *et al*., 2011; Wham *et al*., 2010). Também se tem assistido a um contínuo esforço de harmonização das práticas do aconselhamento genético, acreditação profissional e equidade no acesso aos serviços (Cordier, Lambert, Voelckel, Hosterey-Ugander, & Skirton, 2012).

A nível nacional, o primeiro curso de especialização científica e profissional em aconselhamento genético, que formou e acreditou profissionais, terminou em 2011.

Metcalfe, Aitken e Gaff (2008) descreveram um modelo de avaliação de programas de educação em genética com o intuito de potenciar "boas práticas" e o reconhecimento da profissão. Num artigo incluído num número especial do *Journal of Genetic Counseling*, dedicado ao contexto e princípios da educação e formação dos profissionais do aconselhamento genético, as autoras destacam uma tipologia da avaliação de programas cujas premissas são suscetíveis de aplicação noutros contextos. A tipologia inclui três componentes: i) a avaliação formativa designa o levantamento de necessidades quanto ao programa a desenvolver, clarificando objetivos e desenvolvendo o modelo de implementação e avaliação; ii) a avaliação processual refere-se à monitorização da implementação do programa, verificando a adequação dos objetivos à população-alvo; e iii) a avaliação sumativa caracteriza-se pela análise do impacto do programa na população-alvo, especificando impactos a curto e longo-prazo, e requerendo medidas científicas que reflitam os objetivos iniciais do programa.

Neste sentido, o estudo *Are family-oriented interventions in Portuguese genetics services a remote possibility? Professionals' views on a multifamily intervention for cancer susceptibility families* (*cf.* capítulo III, pág. 159), insere-se na perspetiva da avaliação formativa e processual do programa de intervenção multifamiliar. Incide na negociação e planeamento das condições logísticas necessárias à implementação do programa, estabelecimento de uma lógica participativa de validação e levantamento das necessidades relevantes para a população-alvo junto dos profissionais de saúde (Metcalfe *et al*., 2008). Os estudos do capítulo III contribuem para a literatura ao envolver os profissionais na avaliação de uma intervenção e a refletirem acerca das necessidades com que os serviços se deparam na provisão de apoio psicossocial cada vez mais efetivo aos consultandos e suas famílias.

Os serviços de aconselhamento oncogenético possuem uma orientação predominantemente biomédica. Esta evidência é suportada pela escassez de recursos humanos (sobretudo psicólogos, técnicos de serviço social ou terapeutas familiares), bem como pela prática do aconselhamento genético reportada pelos profissionais, ancorada no modelo de ensino, cujo pendor se centra na provisão de informação (Kessler, 2000). Adicionalmente, as necessidades destacadas pelos profissionais para melhorarem a sua prática incidiram em aspetos conotados com uma orientação psicossocial, como as competências de comunicação e apoio à tomada de decisão.

Embora exploratórios e sem abarcar a realidade nacional dos serviços, os estudos do capítulo III apontam para a existência de carências estruturais (como limitações de financiamento das instituições públicas, de espaço físico e recursos humanos) que obstam à disponibilização de apoio psicossocial de forma consistente e articulada. A definição de um modelo geral de provisão de serviços que contemple coordenação interprofissional e intersectorial e adequada formação dos

profissionais de saúde, de modo extensível aos serviços de saúde primários, foram aspetos organizacionais igualmente destacados. Assim, importa realçar que o apoio psicossocial deverá ser perspetivado de modo coordenado e integrador de vários serviços, especialidades e profissionais que atuam na órbita dos cuidados oncológicos e não enquanto responsabilidade "exclusiva" do aconselhamento genético *per se*. Estas premissas sobressaíram num estudo recente de exploração das práticas clínicas do aconselhamento oncogenético junto de elementos do *Practice Issues Subcommittee of the NSGC Familial Cancer Risk Special Interest Group (Cancer SIG)* (Wham *et al*., 2010). Para além de historicamente o foco da prestação de cuidados em genética e em oncologia divergir, os avanços na epidemiologia genética do cancro representam, para ambas as especialidades médicas, uma mudança paradigmática na prestação dos cuidados. A reorganização dos serviços de saúde oncogenéticos de acordo com os desenvolvimentos da medicina pós-genómica envolve desafios ligados à reconfiguração e redistribuição de papéis profissionais no sentido da interdisciplinaridade entre serviços primários, secundários e terciários (Battista *et al*., 2011; Eeles *et al*., 2007).

Nesta perspetiva insere-se também a necessidade do efetivo envolvimento de estruturas comunitárias na provisão de apoio psicossocial, sobretudo enquanto elo de ligação entre os serviços de saúde primários e terciários e no acompanhamento pós-teste. Estes dados são consistentes com estudos de revisão da literatura recentes sobre modelos de prática e organização de serviços de aconselhamento genético (Battista *et al*., 2011; Cohen, McIlvried, & Schnieders, 2009; Scheuner, Sieverding, & Shekelle, 2008). Abrangendo estudos em países europeus, norte-americanos e Austrália, esses estudos identificaram modelos emergentes na provisão dos cuidados de saúde pós-genómicos, evidenciando a centralidade das abordagens colaborativas entre os múltiplos profissionais de saúde e o envolvimento de profissionais da área psicossocial com formação específica em genética psicossocial. A necessidade de estender a formação sobre genética aos serviços de saúde primária é um aspeto amplamente reconhecido na literatura, nomeadamente quanto à identificação de casos através da interpretação da história familiar, critérios de referenciação para aconselhamento genético, entrevista clínica, dilemas éticos, impactos psicossociais e comunicação da informação genética (Burke *et al*., 2009; Houwink *et al*., 2011; Julian-Reynier *et al*., 2008; Martin & Wilikofsky, 2004). Apesar de algumas iniciativas educativas terem sido delineadas (Carroll *et al*., 2011; Challen *et al*., 2005), os profissionais mencionam necessidades claras que apontam para um contínuo esforço da sua formação, incluindo o desenvolvimento de estratégias efetivas que aumentem as suas competências específicas na área (Nippert *et al*., 2011).

Os constrangimentos estruturais destacados deverão ser entendidos numa perspetiva mais ampla, à luz de lacunas no planeamento da provisão dos serviços. Consideramos que estes aspetos refletem: a inexistência de "tradição psicossocial" nas instituições de saúde; e a tendência centralizadora das

políticas no sentido do escasso envolvimento comunitário. Estes fatores emergem como dados epidemiológicos apontadas na avaliação ao Plano Nacional de Saúde 2004-2010 (WHO, 2011), documento que refere ainda a escassez de oportunidades ao envolvimento proactivo do público e da comunidade, recomendando a inclusão das experiências dos utentes na avaliação, planeamento e implementação de intervenções.

## 4. LIMITAÇÕES E PERSPETIVAS DE PESQUISA

Esta é uma investigação-ação exploratória que visa conhecer um tema pouco estudado, pelo que mais pesquisa neste âmbito é necessária para aprofundar os dados e colmatar as limitações que derivam do desenho metodológico e do enfoque da investigação. Às limitações já mencionadas nos estudos que integram os diferentes capítulos, juntam-se outras emergentes da reflexão da investigação "total" cuja consideração abre novas perspetivas de pesquisa futura.

A investigação não contemplou critérios sociodemográficos específicos nas amostras dos vários estudos, que abarcaram participantes oriundos de diferentes níveis socioculturais e socioeconómicos. É crível que diferenças quanto ao estatuto socioeconómico, grau de escolaridade ou proveniência (contexto urbano, peri-urbano ou rural) tenham impacto diferenciado nas perceções da experiência do aconselhamento oncogenético, no confronto com o risco e com a doença oncológica e nos significados elaborados e socialmente partilhados. Os estudos incluiram apenas participantes europeus-caucasianos, ainda que a literatura enfatize a importância de uma perspetiva multicultural no estudo da experiência do aconselhamento genético, nomeadamente, junto de indivíduos e famílias oriundos de grupos étnicos minoritários (Eeles, Purland, Maher, & Evans, 2007; Lewis, 2002). Consideramos que as questões mencionadas encerram simultaneamente vantagens (semelhança com a população geral que acede a estes serviços, estabelecendo uma relação mais próxima da *realidade*) e desvantagens (negligencia uma análise inclusiva transcultural e perpetua modelos etnocêntricos de interpretação da ciência) (Kleinman, Eisenberg, & Good, 1978).

### 4.1. Alargamento à perspetiva desenvolvimental

Embora a conceptualização exploratória desta investigação inclua uma dimensão desenvolvimental, concebendo o aconselhamento genético a partir da sua justaposição com as fases do ciclo de vida familiar, consideramos que uma perspetiva de pesquisa, potencialmente frutífera, envolveria explorar o funcionamento familiar no confronto com os marcos processuais da experiência do aconselhamento oncogenético (*cf*. Fig. 7.1., pág. 206) em diferentes etapas do ciclo vital da família.

Rolland (2006) delineou um trabalho compreensivo sobre este tópico, atualizando o conceito de perda antecipatória à luz das fases temporais das doenças genéticas e acoplando-o à dimensão evolutiva do ciclo de vida familiar.

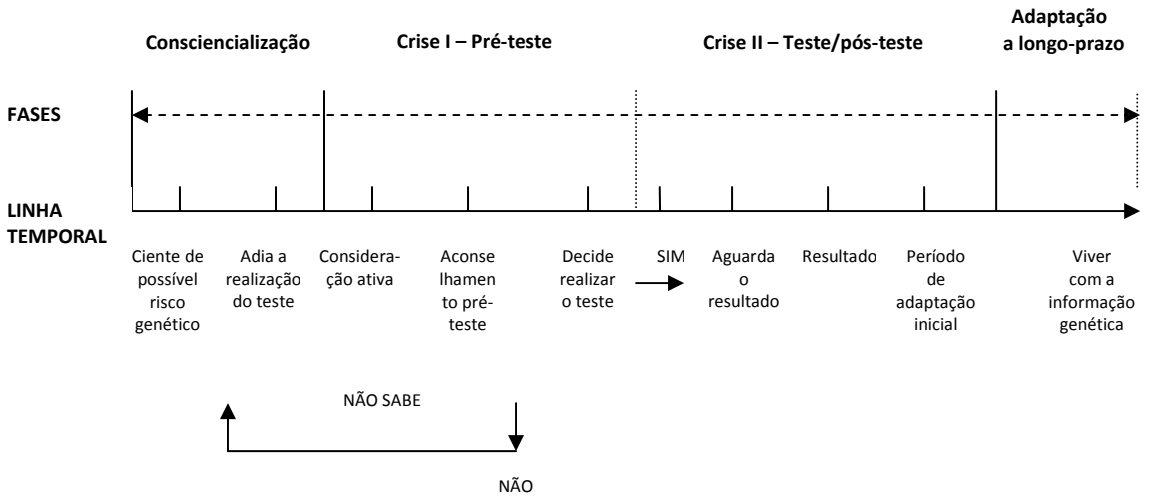
A exploração das experiências com o aconselhamento genético em famílias com configurações distintas da família nuclear tradicional (as "novas formas de família", Relvas & Alarcão, 2002) insere-se nesta perspetiva. Por esta designação entende-se os contextos familiares diversos da família nuclear intacta tradicional, sendo alguns exemplos a família de educador único ou as famílias adotivas. No contexto do aconselhamento genético, esta questão assume relevância dada a crescente diversidade e complexidade de formas de *ser* família para além dos laços estritamente biogenéticos (Richards, 1996). A utilização de tecnologias de reprodução medicamente assistida é um exemplo de tal complexidade, permitindo novas disjunções entre relações biológicas e parentesco (Feetham & Thomson, 2006).

Assim, destacamos como áreas de pesquisa no espectro desenvolvimental: i) a negociação e posterior ajustamento (individual, conjugal e familiar) face à utilização de tecnologias de procriação medicamente assistida, concretamente com o diagnóstico genético pré-implantatório (Werner-Lin *et al*., 2011) e as suas implicações nas relações com a rede social (Gameiro, Boivin, Canavarro, Moura-Ramos, & Soares, 2010); ii) o envolvimento familiar face à realização de testes de suscetibilidade genética em crianças (Clarke, 2010); iii) os significados atribuídos ao risco oncogenético em jovens adultos em diferentes subgrupos (por exemplo, indivíduos com e sem filhos, com e sem companheiro(a), na senda do que Werner-Lin, 2008a, apelidou de "*compressed life-cycle*"), podendo igualmente ser analisada a influência do risco genético em relações de namoro (Klitzman & Sweeney, 2011); e iv) as implicações do risco genético no sentimento de integridade do ego e familiar em pessoas idosas (King & Wynne, 2004), explorando assim a dimensão biológica do legado ou herança (Hunter & Rowles, 2005; Patrão & Sousa, 2010).

### 4.2. Aprofundamento do estudo da adaptação psicossocial ao longo da linha temporal do aconselhamento genético

Pesquisas futuras poderão replicar os estudos integrantes dos capítulos I e II junto de indivíduos que se encontrem nas duas etapas iniciais da fase pré-teste de suscetibilidade, embora o estudo *Experiencing genetic counselling for hereditary cancers: the client's perspetive* (*cf.* capítulo I, pág. 69) tenha incluído dois elementos nesta fase. Assim, seria possível aprofundar o conhecimento de outros aspetos do impacto do aconselhamento oncogenético nos processos de adaptação individual e familiar e, de modo análogo a esta tese, desenvolver intervenções de apoio psicossocial dirigidas às necessidades aí identificadas.

**Fig. 7.2. Fases temporais assintomáticas das doenças genéticas e processo de aconselhamento genético** (adaptado de Rolland & Williams, 2005: 12)




O capítulo II desta tese focou o desenvolvimento de uma intervenção psicoeducativa, dirigida a famílias com suscetibilidade oncogenética acrescida para cancro da mama e ovários e cancro colorectal, isto é, recrutadas através de um consultando portador de mutação(ões) genética(s) deletérias. Apesar de a pesquisa incidir em momentos particulares da linha temporal do aconselhamento oncogenético e respetiva adaptação individual e familiar, não contemplou duas possibilidades que, pela sua frequência e ressonância psicossocial (Ardern-Jones, Kenen, Lynch, Doherty, & Eeles, 2010; Bakos *et al*., 2008), seria pertinente aprofundar: indivíduos (e suas famílias) cujo resultado no teste de suscetibilidade genética foi negativo ou inconclusivo. Contudo, o estudo sobre a experiência familiar do aconselhamento oncogenético (*cf*. capítulo I, pág. 83) incluiu a presença de duas famílias com elementos nestas circunstâncias. Nesse estudo, o uso do genograma na fase inicial da entrevista permitiu verificar a existência de vários elementos na rede familiar com resultado negativo no teste genético. Em geral, a ausência desses indivíduos das entrevistas e da participação nos programas de intervenção multifamiliar implementados sugere a existência de uma perceção familiar que tende a "excluir" tais indivíduos das "conversações sobre o risco". Várias hipóteses compreensivas podem ser delineadas: no estudo de Bakos *e* colaboradores (2008) foram reportadas dificuldades de relacionamento interpessoal entre portadores e não-portadores, com os últimos a revelarem sentimentos de culpa e os primeiros a manifestarem o que os autores descrevem como "ciúme invertido" ("*inverse jealousy"*), traduzindo o desejo de terem na família elementos igualmente portadores para partilhar a sua experiência e identificar-se emocionalmente. Os consultandos com resultados inconclusivos revelam dificuldades

quanto à decisão de envolvimento em intervenções profiláticas de redução de risco (Ardern-Jones, Kenen, Lynch, Doherty, & Eeles, 2010). Estes dados sugerem a pertinência em incorporar, nas abordagens familiares ao estudo do risco oncogenético, a perspetiva de elementos com resultado positivo, negativo e inconclusivo ao teste genético.

### 4.3. Apoio à comunicação intrafamiliar sobre a informação genética

O aprofundamento dos perfis de transmissão da informação genética e a caracterização dos seus intervenientes constitui um relevante recurso para o desenvolvimento de intervenções de apoio, pois trata-se de um processo complexo com implicações psicossociais e no acesso a cuidados de saúde. A literatura evidencia que os profissionais tendem a encorajar a transmissão de informação sobre o risco genético a nível intrafamiliar, sem contudo disponibilizarem apoio específico neste processo (Forrest, Delatycki, Curnow, Skene, & Aitken, 2010). Além disso, fatores emocionais, relacionais, geográficos ou especificamente comunicacionais concorrem para a disseminação da informação genética na família (Foster, Eeles, Ardern-Jones, Moynihan, & Watson, 2004; Mesters, Ausems, Eichhorn, & Vasen, 2005; Vos *et al*., 2011). Estas circunstâncias obstam a que vários elementos potencialmente em risco tenham acesso a informação genética suscetível de auxiliar possibilidades pré-sintomáticas de gestão da saúde. Resulta daqui a necessidade de mais estudos conducentes à clarificação de estratégias adequadas para assistir os consultandos com soluções dirigidas às suas idiossincrasias individuais, relacionais e familiares na transmissão de informação genética.

Apesar da utilidade prática inerente à disponibilização de informação genética relevante para a gestão da saúde individual e familiar, e para a promoção de decisões informadas, subjazem questões de natureza prática e ética que têm merecido aturada reflexão na literatura. O debate centra-se nas questões da privacidade da informação genética e o contacto com sujeitos que não são clientes / utentes do serviço (i.e., os familiares do probando ou caso-index); e as eventuais repercussões adversas a nível individual e familiar decorrentes de sentimentos de pressão para o envolvimento em testes genéticos. Suthers, Armstrong, McCormack e Trott (2006) argumentam que a provisão deste serviço de apoio por uma entidade mediadora "externa" possibilita aos familiares a separação das componentes emocional e instrumental. Esta assunção decorre dos resultados obtidos pelos referidos autores, indicando que o envio de cartas-resumo com informação genética aos familiares potencialmente em risco, fomentou maior procura de aconselhamento genético, da parte destes, nos dois anos subsequentes, comparativamente com o grupo de controlo, isto é, a "gestão familiar" da transmissão da informação genética (40% e 23%, respetivamente); ainda se verificou que, no grupo da intervenção, o subgrupo de familiares notificados por carta enviada pelo serviço de genética, recorreram ao aconselhamento genético em maior proporção do

que o subgrupo a quem a carta foi entregue pelo probando (46% e 24%, respetivamente). Outros estudos, porém, salientam que os indivíduos encaram como sendo sua responsabilidade a disseminação da informação genética aos familiares, apesar de reportarem necessidades de apoio neste processo (Gaff *et al*., 2007). Estes dados sugerem a necessidade de apoio para além das estratégias meramente informativas (como o uso de cartas a serem distribuídas pelos consultandos aos familiares identificados como estando potencialmente em risco). Algumas sugestões sobre como os profissionais poderão auxiliar os consultandos neste processo foram avançadas, embora escasseiem estudos documentando os seus efeitos na capacitação comunicacional dos consultandos (Gaff *et al*., 2007). Por outro lado, uma abordagen ecológica-sistémica ao tema proporcionaria um conhecimento mais aprofundado dos processos individuais, familiares e comunitários envolvidos, permitindo ultrapassar medidas reducionistas que tendem a universalizar soluções para um processo multideterminado e que envolve mecanismos decisórios de extrema complexidade. Consideramos que uma abordagem potencialmente frutífera neste domínio, e eticamente mais fiável, inclui o aprofundamento da natureza variável das barreiras à comunicação intrafamiliar, de modo a desenvolver formas de apoio distintas e adequadas às especificidades das famílias. Neste ponto, a categorização proposta por Forrest e colaboradores (2003) parece-nos particularmente útil, pois os autores sugerem que as hesitações comunicacionais na família podem assumir conotações positivas (actuam em benefício de alguém, *e.g*., protecção face aos potenciais efeitos nefastos do acesso à informação), negativas (incapacidade para agir ou ultrapassar dificuldades) ou neutras (em que existe a percepção de que não é necessário apoio externo) (Forrest *et al*., 2003).

Com efeito, consideramos que um dos desafios mais prementes nos cuidados de saúde da era (pós)genómica envolve o desenvolvimento de intervenções de apoio à comunicação da informação genética na família, passíveis de contemplarem a sua efetiva disseminação, e respeitando as idiossincrasias relacionaise princípios ético-morais dos indivíduos e familiares.

Por outro lado, a inclusão de temas alternativos nas sessões da intervenção multifamiliar foi destacada no cruzamento qualitativo dos dados da avaliação do programa, incluindo: formas alternativas de abordar a gestão do *stress* e identidade familiar, ou apoio à comunicação intrafamiliar na transmissão da informação genética.

Existem intervenções específicas quanto à comunicação intrafamiliar da informação genética (Daly *et al*., 1999), e a literatura identifica-o como uma das necessidades mais prementes, indicando a sua presença em intervenções de caráter psicoeducativo, inclusive no programa que serviu de base ao desenvolvido nesta tese (Chiquelho, Neves, Mendes, Relvas, & Sousa, 2011; Sousa, Mendes, & Relvas, 2007). O tema não foi incluído porque: não foi mencionado nas entrevistas pré-teste de levantamento de necessidades; e as premissas psicoeducativas enfatizam a educação para a saúde,

que apenas indiretamente foca a facilitação de canais de comunicação de maior fluidez intra- e interfamílias. O acesso à componente informativa e à partilha de experiências com as outras famílias, acrescido da facilitação de uma atmosfera "normalizadora" de sentimentos e do discurso sobre a informação genética, contribui, ainda que indiretamente, para melhorar a capacidade de transmissão da informação genética intrafamiliar. De facto, traduz-se num recurso efetivo à compreensão da informação genética que estimula o esclarecimento de dúvidas existentes entre os membros da família (Plumridge *et al*., 2011). Todavia, assumimos agora que o apoio à transmissão da informação genética na família seria um importante tópico a incluir em futuros programas de intervenção.

### 4.4. Estudos de eficácia: dados quantitativos, grupos de controlo e significado clínico

O programa de intervenção multifamiliar apresenta limitações que sugerem a necessidade de considerar alternativas quanto a novos percursos metodológicos e à sua estrutura e conteúdos. A triangulação dos resultados da avaliação do programa de intervenção (participantes e profissionais) representa um esforço no sentido da sua validação e fiabilização.

O recurso exclusivo a metodologias qualitativas para analisar o impacto psicossocial da intervenção nos participantes e o seu caráter não-experimental merece ser considerado. A ausência de uma componente avaliativa sumativa limita a validade do programa, pelo que a sua exploração abre uma perspetiva de pesquisa futura, concretamente quanto à avaliação dos impactos psicossociais a curto e longo-prazo. O uso combinado de métodos de recolha de dados quantitativos e qualitativos (triangulação de métodos[37]) permitiria, potencialmente, uma avaliação mais precisa do impacto da intervenção quanto a fatores específicos da adaptação psicossocial, a nível individual e familiar, de acordo com a argumentação de Kasparian, Wakefield e Meiser (2007: 706):

> "*Importantly, while there are both process studies and outcome studies, a final overarching goal in genetic counselling research is to link processes to outcomes as a way of demonstrating the effectiveness of counselling practice. In order to do so, researchers must be familiar with the many usable tools available for the evaluation of psychosocial adaptation to genetic information*".

[37] Patton (1990) refere-se à triangulação como um importante recurso no incremento de qualidade e credibilidade da análise qualitativa, enumerando quatro tipos: i) verificar a consistência dos resultados obtidos através de diferentes métodos de recolha de dados (triangulação de métodos); ii) testar a consistência de diferentes fontes de dados (triangulação de fontes); iii) recorrendo a múltiplos avaliadores para rever os resultados (triangulação da análise); e iv) utilizando várias perspetivas ou teorias para interpretar os dados (triangulação de teorias/perspetivas). O estudo *Are family-oriented interventions in Portuguese genetics services a remote possibility? Professionals' views on a multifamily intervention for cancer susceptibility families* (*cf.* capítulo III, pág. 159) procurou integrar este requisito através da triangulação da análise.

Contudo, o objetivo orientador do programa de intervenção centrou-se na verificação da adequação estrutural e temática do seu formato e da sua viabilidade enquanto recurso psicossocial de apoio às famílias em risco oncogenético. Os participantes nos dois grupos reportaram a adequação da estrutura e conteúdos do programa às suas necessidades, pelo que a inclusão das medidas quantitativas específicas pré- e pós-teste (por exemplo, recorrendo a grupos aleatórios) apresenta-se como uma perspetiva de pesquisa futura pertinente para aprofundar o tipo de benefícios psicossociais e sua extensão. Chiquelho, Neves, Mendes, Relvas e Sousa (2011) descreveram um estudo quasi-experimental longitudinal com o *ProFamílias*, o programa psicoeducativo multifamiliar que serviu de base ao incluído nos estudos do capítulo II, dirigido a doentes oncológios e suas famílias a experienciarem a fase de crise da doença (até 6 meses após o diagnóstico, *cf*. Rolland, 1994). Nesse estudo, foram comparadas duas subamostras (grupo experimental e controlo) em momentos distintos (pré-intervenção e pós- intervenção, logo após a participação no programa e um ano após) quanto a: ajustamento psicossocial, funcionamento familiar e *stress* percebido. Os dados permitiram iluminar o trajeto do impacto psicossocial das intervenções multifamiliares, apontando diferenças significativas entre os dois grupos quanto ao ajustamento psicossocial dos pacientes e aos níveis de coesão e adaptabilidade familiar (mais elevados nos grupos controlo), enquanto que as diferenças nos níveis de *stress* percebido não se revelaram significativas.

Kasparian e colaboradores (2007) elencaram um conjunto de escalas de avaliação de medidas psicossociais comummente utilizadas em pesquisas no âmbito do aconselhamento genético, agrupando-as em categorias temáticas. As mais pertinentes neste contexto seriam: expectativas e crenças; compreensão da informação genética; ajustamento psicológico; tomada de decisão; e funcionamento familiar. Na tabela 7.3. elencam-se algumas escalas de avaliação para cada categoria mencionada.

**Tabela 7.3. Escalas de avaliação[38] de medidas psicossociais usadas em investigação no aconselhamento genético** (adaptado de Kasparian *et al*., 2007)

| Categoria temática | Escala | Autores |
|---|---|---|
| Expectativas e crenças | *Background, Needs and Expectations for Genetic Counseling Scale* | Peters & Petrill (2010) |
| Compreensão da informação genética | *Breast Cancer and Heredity Knowledge Scale* | Ondrusek *et al.* (1999) |
| | *Breast Cancer Genetic Counselling Knowledge* | Erblich *et al.* (2005) |
| Ajustamento psicológico | *Psychological Adaptation to Genetic Information Scale* | Read, Perry, & Duffy (2005) |

[38] As escalas apresentadas revelam características variáveis quanto às propriedades psicométricas (consistência interna, fiabilidade teste-reteste e validade de constructo) (Kasparian *et al*., 2007). A validação destas escalas para a população portuguesa seria outra perspetiva de pesquisa que consideramos pertinente.

| | | |
|---|---|---|
| | *Multidimensional Impact of Cancer Risk* | Cella *et al*. (2002) |
| | *Genetic Testing Psychosocial Risk Screening Tool* | Esplen *et al*. (2011) |
| | *Brief COPE* | Carver (1997) |
| Tomada de decisão | *Multidimensional Measure of Informed Choice* | Michie *et al*. (2002) |
| | *Life Orientation Test-Oriented* | Scheier *et al*. (1994) |
| | *Decisional Conflict Scale* | O'Connor (1995) |
| | *Decision Evaluation Scale* | Stalmeier *et al*. (2005) |
| Funcionamento familiar | *Openness to Discuss Cancer in the Family Scale* | Mesters et al. (1997) |
| | *Family Communication Questionnaire* | Hughes *et al*. (2002) |
| | *Family Assessment Measure* | Cappelli *et al*. (2005) |

**Tabela 7.3. Escalas de avaliação de medidas psicossociais usadas em investigação no aconselhamento genético** (adaptado de Kasparian *et al*., 2007) (cont.)

Consideramos que a utilização exclusiva de medidas apriorísticas para aferir o impacto de intervenções descura outros níveis de análise e dimensões da experiência suscetíveis de serem valorizados pelos participantes e desempenharem uma função relevante na adaptação psicossocial (Mirin & Namerow, 1991). O enfoque destas escalas está tipicamente conotado com objetivos pré-determinados e índices de funcionamento normativos (como redução, eliminação ou mitigação de sintomatologia específica). Este processo de avaliação tende a não contemplar processos de mudança ou aspetos eventualmente emergentes durante a intervenção que não os definidos previamente; assim, não permite capturar aspetos do funcionamento psicossocial com significado clínico e relevância prática para o indivíduo e outros significativos (Kazdin, 1999).

### 4.5. Da flexibilização da intervenção à diversificação do apoio psicossocial

Em geral, o número de sessões, duração e periodicidade foram apontados como adequados, embora alguns participantes tenham indicado alterações (aumento da duração das sessões e a inclusão de outros temas). A postura dos técnicos foi percebida como adequada, apesar de maior directividade na gestão do envolvimento dos participantes nos temas a abordar nas sessões ter sido sugerido.

Uma limitação evidente à implementação do programa é amplamente referida na literatura: as dificuldades no recrutamento dos participantes (Gonzalez & Steinglass, 2002; McAllister, 2005; Ostroff, Ross, Steinglass, Ronis-Tobin, & Singh, 2004). Uma das formas que encontrámos para contornar este obstáculo foi o recurso à seleção de amostras de conveniência, usando o critério da proximidade geográfica ao local de realização das sessões, procurando obviar constrangimentos financeiros, temporais e logísticos. A utilização deste critério deveu-se à experiência prévia do investigador no desenvolvimento e implementação de intervenções multifamiliares em contexto hospitalar. As restrições no recrutamento dificultam a universalização da intervenção, deixando antever obstáculos à sua efetiva incorporação em instituições de saúde.

A versão "condensada" do programa num *workshop* multifamiliar de 1 dia com sessões mensais de seguimento apresenta-se como uma alternativa interessante face às restrições ao envolvimento das famílias num formato mais intensivo (4 sessões semanais). O caráter esporádico desta modalidade suscita, no entanto, dúvidas face ao impacto na efetiva gestão psicossocial das famílias, embora bons resultados tenham sido apresentados numa intervenção em regiões rurais remotas de Vermont, nos E.U.A. (McKinnon, Naud, Ashikaga, Colletti, & Wood, 2007) e em contexto hospitalar (Steinglass, Ostroff, & Steinglass, 2011). Adicionalmente, a possibilidade de encetar estudos comparativos quanto aos níveis de adaptação psicossocial e funcionamento familiar obtidos na participação no programa apresentado e na versão *workshop*, permitiria a obtenção de dados úteis quanto à planificação da utilização de ambas as modalidades.

Tipicamente as famílias que se envolvem em intervenções multifamiliares revelam elevados níveis de coesão (Chiquelho *et al*., 2011; Gonzalez & Steinglass, 2002), donde resulta a necessidade de planear e desenvolver intervenções acessíveis a núcleos familiares menos coesos. Para abarcar o caráter crónico e variável das necessidades psicossociais de indivíduos e famílias, são necessárias intervenções dirigidas a dificuldades específicas previamente identificadas e em formatos diversificados (designadamente, individualizados ou unifamiliares); a facilitação de grupos de apoio em contexto comunitário merece igualmente ser considerado. O formato multifamiliar não representa um meio de apoio universalmente aceite; ou seja, nem todos os indivíduos e famílias estão disponíveis para integrarem grupos de discussão multifamílias no contexto do risco genético. As desvantagens ou razões para a não participação nestes grupos são: a impossibilidade em garantir o anonimato e a confidencialidade, e a partilha de experiências do foro íntimo com elevado potencial de ativação emocional em situações face-a-face com estranhos (Plumridge *et al*., 2011).

### 4.6. Mais pesquisa sobre a provisão de serviços no aconselhamento oncogenético

Os estudos do capítulo III reúnem uma base exploratória para equacionar os desafios mencionados numa perspetiva nacional. Porém, mais estudos devotados à provisão de aconselhamento oncogenético em Portugal (e a nível internacional) são necessários, nomeadamente quanto ao planeamento de serviços multidisciplinares integrados com os cuidados de saúde primários, considerando as preocupações que os testes de suscetibilidade genética para doenças comuns encerram numa perspetiva de saúde pública (Van El, Cornell, & ESHG Public and Professional Public Committee, 2011). Estabelecer uma base de dados nacional sobre a provisão do aconselhamento genético, profissionais envolvidos, procedimentos e modelos de prática, e necessidades específicas a nível educativo e organizacional, é uma perspetiva de pesquisa necessária.

O recurso a *focus group* com uma amostra mais alargada e diversificada de profissionais, peritos e decisores políticos seria uma opção a considerar; noutros estudos com profissionais, o

preenchimento de questionários via *Internet* tem sido uma metodologia frequente. Neste ponto, um enfoque regional (envolvendo representantes das Direções Regionais de Saúde) seria porventura mais exequível, com a vantagem de poder aquilatar idiossincrasias locais de modo mais preciso, por exemplo, especificar informadores-chave para os vários componentes do aconselhamento genético, com diferentes percursos, experiência e formação. Envolver elementos da comunidade e representantes de associações de doentes e de familiares de doentes no levantamento de necessidades psicossociais é igualmente pertinente, na senda de um esforço integrador e orientador do planeamento da provisão de serviços.

As perspetivas de pesquisa elencadas assumem relevância acrescida considerando a recente conclusão da formação de profissionais de aconselhamento genético devidamente acreditados. Explorar junto de uma base alargada de representantes-chave na provisão dos serviços de genética as suas expectativas face à inclusão daqueles profissionais nos serviços, é outra perspetiva de pesquisa potencialmente útil.

## 5. RECOMENDAÇÕES PARA SERVIÇOS PSICOSSOCIAIS NO ACONSELHAMENTO ONCOGENÉTICO

Esta investigação reafirma a pertinência de uma abordagem centrada na família no aconselhamento genético (McDaniel, 2005; Miller, McDaniel, Rolland & Feetham, 2006; Weil, 2000). Salienta também a relevância do desenvolvimento de formas diversificadas de apoio psicossocial ao longo do processo de aconselhamento oncogenético, que contemplem a dimensão evolutiva do *continuum* biopsicossocial que inclui indivíduo, família, rede social e comunidade (Engel, 1977).

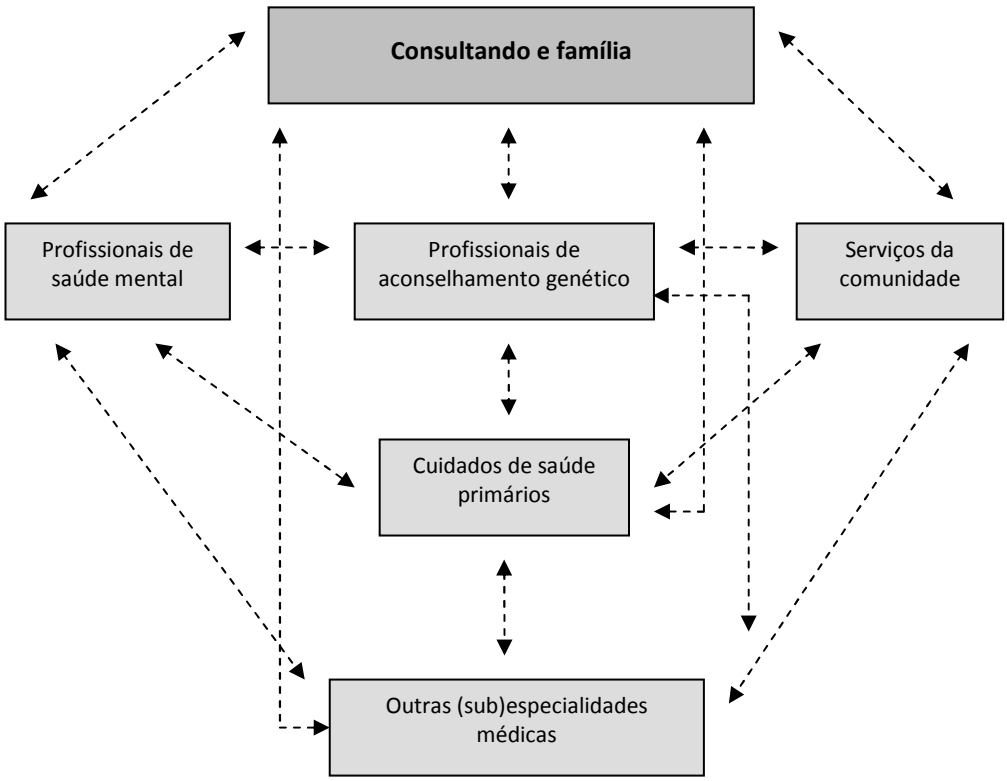
Seguindo a premissa que concebe a interdependência entre os dados científicos e saúde pública (Almeida, 2010), e com base nos resultados desta investigação e na pesquisa da literatura, proporemos de seguida algumas recomendações para a provisão de serviços psicossociais no âmbito do aconselhamento oncogenético. Pretende-se que possam contribuir para a discussão de temas associados ao planeamento e implementação de serviços neste contexto.

A literatura refere de modo consistente dados e aplicações clínicas que destacam um enfoque psicossocial e familiar no contexto do aconselhamento (onco)genético (Eunupu, 1997; Kenen, Ardern-Jones & Eeles, 2006; Kenen & Peters, 2001; Koehly *et al*., 2003; Speice, McDaniel, Rowley, & Loader, 2002; Werner-Lin, 2007; 2008); algumas das mais relevantes associações de profissionais e redes de excelência enfatizam a necessidade de os cuidados de saúde genómicos incluírem um enfoque familiar e psicossocial (Fryer & Cheese, 1998; Genetic Alliance, 2001, Kääriäinen *et al*., 2010). Na esteira do que vários autores há muito preconizam (Kessler, 1979; Reed, 1955 *in* Resta, 2006), inclusive entre nós (Zagalo-Cardoso, 1989), alguns modelos

conceptuais foram propostos para a avaliação, compreensão e intervenção psicossocial no aconselhamento genético (Etchegary, Lemyre, & Wilson, 2010; Rolland & Williams, 2005; Soldan, Street, Gray, Binedell, & Harper, 2000; Street & Soldan, 1998; Tluczek *et al*., 2010). É também conhecida a relação positiva entre bem-estar psicossocial e a saúde física (Parkes, 1972), bem como a influência recíproca da dinâmica familiar e comportamentos face à doença, adesão a tratamentos e curso dos sintomas (Campblell, 2003; Weihs, Fisher, & Baird, 2002), incluindo o contexto oncogenético (Rees, Fry, & Cull, 2001).

Retome-se a linha temporal do aconselhamento genético na fase pré-doença, anterior à manifestação clínica da doença (Rolland & Williams, 2005; *cf*. capítulo I, pág. 70 e Fig.7.2., pág. 216). Segundo os autores, o seu uso combinado com a tipologia "clássica" das fases da história natural das doenças crónicas (Rolland, 1994), providencia um modelo de avaliação e intervenção nas doenças genéticas numa abordagem individual, familiar e desenvolvimental. Consideramos a referida tipologia e o quadro conceptual apresentado nas conclusões gerais desta tese (*cf*. Fig. 7.1., pág. 206) como grelhas de referência para as recomendações que apresentamos (Figura 7.4).

**Fig. 7.4. Provisão de serviços no aconselhamento oncogenético** (adaptado de McDaniel, Peters, & Acheson, 2006: 531)



Por conseguinte, o modelo fundacional dos serviços oncogenéticos requer coordenação interprofissional, pressupondo um cariz multidisciplinar e colaborativo na provisão dos cuidados. Dois princípios orientadores norteiam as nossas recomendações: i) abordagem centrada no paciente e na família, visando assegurar o seu bem-estar e reconhecendo-os como parceiros na avaliação, planeamento, provisão e avaliação dos cuidados de saúde (Adler & Page, 2008; Mead & Bower, 2000); e ii) necessidade de formação adequada em genética psicossocial dos vários profissionais envolvidos, em particular quanto à adaptação familiar ao risco genético. Assim, considerando três componentes (formação dos profissionais, organização dos serviços e ligação aos cuidados de saúde primários e comunidade, e acompanhamento a consultandos e famílias), recomenda-se:

### 5.1. Formação dos profissionais

- Incluir uma componente sobre a adaptação psicossocial e familiar ao aconselhamento e risco oncogenético nos programas de formação graduada dos profissionais de aconselhamento genético[39] e outros profissionais de saúde envolvidos na prestação de cuidados neste âmbito (geneticistas, enfermeiro(a)s, oncologistas, psicólogo(a)s, técnico(a)s de serviço social).
- Incluir uma componente de conceitos básicos de genética aplicados ao contexto do aconselhamento genético nos currículos de formação da especialidade de medicina geral e familiar; os internatos desta especialidade, bem como de oncologia, poderão contemplar a opção de estágios em serviços de (onco)genética envolvendo a observação e acompanhamento dos procedimentos do aconselhamento genético.
- Promover a formação dos profissionais quanto a questões emergentes da adaptação psicossocial ao risco e aconselhamento oncogenético, incluindo as heurísticas mais utilizadas por indivíduos e famílias na gestão subjetiva do risco (Kenen, Ardern-Jones, & Eeles, 2003).
- Desenvolver ações de sensibilização e formação em aspetos relevantes para o aconselhamento genético, dirigidas a médicos de família e outros profissionais dos cuidados de saúde primários, contemplando: conceitos básicos, familiarização com o desenho do *pedigree*, critérios de Amesterdão e interpretação da história médica familiar, comunicação de informação genética, e impactos psicossociais. Para tal, o envolvimento de profissionais do aconselhamento genético seria relevante, através da produção de material informático e didático, incluindo o desenvolvimento de um sítio na *Internet* e a

[39] O Mestrado Profissionalizante em Aconselhamento Genético, do ICBAS, Universidade do Porto, contou, no seu plano curricular, com módulos específicos sobre o impacto psicossocial individual e familiar do risco e aconselhamento genético (Paneque & Sequeiros, 2010).

dinamização de *workshops*[40]; estabelecer uma rede inclusiva de especialistas na área para implementar um levantamento de necessidades e monitorizar o desenvolvimento destas iniciativas.

- Promover a formação e supervisão dos profissionais quanto a questões éticas, tais como, autonomia individual e familiar, direito a (não) saber, privacidade, não-directividade, paternalismo médico-genético, cuidados de saúde multiculturais e com minorias étnicas, enviesamento na apresentação de informação e aconselhamento, e consciencialização do *self* do profissional no encontro clínico.

**5.2. Organização dos serviços e ligação aos cuidados de saúde primários e comunidade**

- Para além dos profissionais médicos, a estrutura técnica dos serviços de oncogenética deve integrar psicólogo(s)s, terapeutas familiares, técnico(a)s de serviço social e profissionais do aconselhamento genético. Esta estrutura deverá ter ligação aos profissionais de outras especialidades envolvidas nos cuidados oncogenéticos, bem como aos cuidados de saúde primários e associações da comunidade.
- Os serviços deverão agilizar a disseminação de material informativo dirigido ao público sobre risco e aconselhamento genético, englobando o impacto psicossocial[41], e sobre o momento e procedimentos para requerer aconselhamento oncogenético (Mackay, Schulz, Rubinelli, & Pithers, 2007). Estas medidas, dirigidas a populações com baixos níveis de literacia, revelaram-se funcionais (Joseph *et al*., 2010; Lubitz *et al*., 2007). O uso de um sítio na *Internet* com informação personalizada, pré-aconselhamento genético, permitiu que os consultandos revelassem perspetivas realistas acerca do processo e diminuição das necessidades informativas na primeira sessão de aconselhamento oncogenético (Albada, van Dulmen, Ausems, Bensing, & Van Dulmen, 2011).
- Equacionar o envolvimento de associações de doentes e familiares de doentes na caracterização das suas necessidades e prioridades de investigação científica, bem como na agilização da provisão de serviços de apoio psicossocial (por exemplo, através da referenciação de famílias para acompanhamento psicossocial). O envolvimento destas

[40] Alguns estudos avaliaram a utilização de ferramentas na educação de profissionais de saúde primários, incluindo: 1) apoio computorizado à interpretação da história familiar (Emery *et al*., 2000); 2) *packs* informativo-didáticos, como o *GenetiKit*, um portfólio que inclui (Carroll *et al*., 2011: 2): "*i) a pen and paper family history tool, ii) a list of genetics 'pearls' (important facts), iii) risk triage and management cards for HBOC and HCRC, iv) a simple aid memoire outlining the possible consequences of genetic test results, and v) informative aids to help patients self-identify their risk of hereditary cancers*"; consultar http://www.mountsinai.on.ca/care/family-medicine-genetics-program/the-genetikit-project, para acesso ao *GenetiKit*); e 3) *workshops* (Carroll, Rideout, & Wilson, 2009). Estas intervenções proporcionaram, de um modo geral, um acréscimo autorreportado de conhecimentos por parte dos profissionais. Alguns sítios na internet ligados a organizações profissionais disponibilizam material informativo (*e.g*. http://www.cfp.ca/search?submit=yes&tocsectionid=Genetics).

[41] http://www.eurogentest.org/blocks/leaflets/pdf/english/cancer.pdf.

associações em parcerias com instituições de cuidados de saúde primários em comunidades rurais distantes dos centros urbanos pode estabelecer-se como um recurso significativo.

### 5.3. Acompanhamento a consultandos e famílias

- Incorporar uma sessão de avaliação psicossocial, prévia à sessão de aconselhamento oncogenético pré-teste, para antecipar dificuldades psicossociais e potenciar recursos individuais e familiares para o processo de aconselhamento oncogenético. Também se deverá informar sobre formas de apoio psicossocial disponíveis para as necessidades específicas identificadas, incluindo o acompanhamento à família; providenciar material informativo acerca das questões médico-genéticas e impacto psicossocial associado aos testes genéticos, a nível individual e na família. Algumas ferramentas podem ser utilizadas na avaliação, tais como: *Background, Needs and Expectations for Genetic Counseling Scale* (Peters & Petrill, 2010), *Genetic Testing Psychosocial Risk Screening Tool* (Esplen *et al*., 2011), *Colored Eco-Genetic Relationship Map* (CEGRM) (Kenen & Peters, 2001; Peters, Hoskins, Prindiville, Kenen, & Greene, 2006), e Instrumento de Análise da Rede Social Pessoal (IARSP-R) (Alarcão & Sousa, 2007).
- Integrar no protocolo de aconselhamento oncogenético, na(s) sessão(ões) de aconselhamento pré-teste, uma breve avaliação do funcionamento familiar. De acordo com as necessidades, direitos e preferências dos consultandos, a inclusão de familiares e outros significativos na sessão de aconselhamento pré-teste deve ser equacionada previamente. Questões acerca da dinâmica familiar, existência de conflitos, padrões de comunicação, expectativas face aos testes genéticos, fontes de *stress* e de suporte, deverão ser englobadas. A utilização do genograma *orientado para a doença* pode ser uma boa opção (Alarcão, 2002).
- Englobar o *Psychological Model for Presymptomatic Test Interviews* (Soldan *et al*., 2000) na(s) sessão(ões) de aconselhamento pré-teste como forma de potenciar a tomada de decisão informada acerca da realização do teste genético. Trata-se de um modelo incorporado no contexto da doença de Huntington e outras doenças neurológicas de início tardio, que inclui as seguintes fases: clarificação, consideração, educação, reflexão e decisão. A utilização de questões centradas na família deve ser encorajada[42].

[42] Sobel e Cowan (2000: 48) propuseram, no contexto do protocolo para o teste pré-sintomático da doença de Huntington, as seguintes questões, passíveis de adaptação ao contexto oncogenético: *How has cancer been dealt within your family? Is testing for cancer susceptibility breaking any family rules? How do you think your testing is viewed by other family members? Who knows you want to do this (testing)? With whom would you share the results (if they are positive or negative)? From whom would you withhold the results, and how would that secret impact your relationship? If you tested positive or negative, how would that affect your relationship with your siblings and with your children?*".

- A apresentação das estimativas de risco aos consultandos deverá envolver descrições verbais e gráficas, incluindo o uso de percentagens e frequências e o recurso a metáforas, quando apropriado (Edwards *et al*., 2008; Henneman *et al*., 2011).
- Incluir no aconselhamento pré ou pós-teste, apoio à transmissão de informação genética a familiares potencialmente em risco (Daly *et al*., 1999).
- Contactar telefonicamente os consultandos durante o período entre a realização do teste genético e o conhecimento dos resultados, disponibilizando apoio psicológico. Considerar a dinamização de grupos psicoeducativos neste hiato temporal dirigidos a indivíduos em risco e suas famílias.
- Na sessão pós-teste de divulgação dos resultados, a disponibilização de formas de apoio psicológico individuais e à família deve ser equacionada, incluindo a participação em grupos de discussão multifamiliares ou outros (sem os familiares); neste tipo de intervenção, poderão ser integrados temas específicos de acordo com as necessidades psicossociais dos participantes, já previamente avaliadas, em módulos / *workshops* psicoeducativos.
- Marcar uma sessão de seguimento para cerca de um mês após a transmissão dos resultados, aberta à inclusão de familiares e outros significativos, com o objetivo de aceder a preocupações e dificuldades psicossociais emergentes. Esta sessão poderá envolver uma exploração da adaptação do sistema familiar à interseção entre a vivência do risco genético e a etapa de desenvolvimento individual e familiar, que poderá redundar em proposta de seguimento psicoterapêutico.
- Debater a incorporação de recursos de videoconferência e telessaúde em articulação com serviços de saúde primários de regiões rurais distantes dos centros urbanos (D'Agincourt-Canning *et al*., 2008).
- Disponibilizar apoios específicos à tomada de decisão quanto ao envolvimento em intervenções profiláticas de redução de risco, incluindo o recurso a materiais informativos, como folhetos (Metcalfe *et al*., 2007), *cd-rom* (Schwartz *et al*., 2009), vídeos (Tiller *et al*., 2006) e através da *Internet* com recurso a *software* específico (Culver *et al*., 2011). Neste âmbito, a promoção de grupos de apoio que incluam indivíduos que já efetuaram cirurgias profiláticas pode funcionar como *fórum* de apoio social, requerendo indivíduos disponíveis (Speice *et al*, 2002).

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